

RARE DISEASE DATA SPECIFICATION

100,000 Genomes Project



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Contents

1	Introduction	4
	This document	4
	Reporting	4
	Classes of Data	4
	Completeness	5
	File upload	6
2	Essential Sample Metadata (12499.3)	7
2.1	Participant Information (12500.3)	7
2.1.1	Participant (12501.3)	7
2.1.2	Contact Details (12528.2)	9
2.1.3	Consent (12541.5)	10
2.1.4	Clinical Information (12553.2)	11
2.1.5	Disease and Eligibility (12579.3)	13
2.1.6	Family (12586.3)	15
2.2	Sample Information (29287.3)	18
2.2.1	Test Results (29288.2)	19
2.2.2	Blood Sample (29089.3)	20
3	Core Data (12736.3)	21
3.1	Participant Identifiers	21
3.2	Pedigree (12772.3)	22
3.2.1	Pedigree Member (33574.2)	22
3.3	Investigation (29497.5)	25
3.3.1	Phenotype Report (12950.3)	26
3.3.2	General Observations (29290.3)	30
3.3.3	Genetic Investigations (12759.3)	33
3.3.4	Biopsy (29498.2)	36
3.3.5	Imaging Diagnostics (29736.2)	41
3.3.6	Laboratory Test Report (29869.2)	53
3.3.7	Non-imaging Diagnostics (30107.2)	131
3.3.8	Patient History (30108.2)	147
3.3.9	Performance Measures (30109.2)	151
3.3.10	Vital signs (30111.2)	155
3.4	Diagnoses (31151.3)	157
3.5	Intervention (29528.2)	157
3.5.1	Medication (29852.1)	157
3.5.2	Surgical Procedure (29856.1)	157
3.5.3	Other Procedure (29859.1)	159
3.6	Withdrawal	160
3.7	Death Details (33514.1)	161
4	Additional Data (12786.1)	162
5	Value Domains	163

1 Introduction

Genomic Medicine Centres are asked to supply detailed patient information to accompany the samples submitted for analysis. The success of the 100,000 Genomes Project – and the quality and value of the clinical reports produced – rests upon the accuracy and completeness of the information supplied.

The information is divided into

- *essential sample metadata* – information about the sample and participant, provided before the sample is sent for sequencing
- *core data* – information on relevant clinical events and observations made at the time of registration or drawn from existing patient records, provided within six weeks of sample collection
- *additional data* – information on subsequent, relevant clinical events and observations, provided within four weeks of the event in question

Which events and observations are deemed relevant will depend upon the rare disease(s) identified when the participant is registered.

Where a participant's condition matches more than one of the listed diseases, Genomic Medicine Centres are asked to report on relevant events and observations for each of the diseases present.

This document

This document describes the different classes of data expected and the datatypes employed and is aimed as a guide to those developing systems for data collection. Each data item is associated with a name, a brief explanation, a multiplicity, and a datatype. In this version of the document, the data item identifiers have been included, to facilitate look-up in the current version of the Data Model Catalogue v1.3.

The relationships between specific disorders and specific data elements are provided in the Data Model Catalogue and the corresponding Rare Disease Data Models v1.3 document.

Reporting

The information required can be provided in two ways: using the web-based case report forms provided by Genomics England, or via a local, integrated records system that can produce reports in XML format for subsequent uploading.

Genomic Medicine Centres are strongly encouraged to develop local, integrated reporting systems for rare disease. As well as providing a valuable resource for patient management and local research activity, such systems will greatly facilitate the process of clinical review prior to submission.

Nevertheless, it is recognised that the wide range of data items required, and the relatively small number of patients involved, may make *comprehensive* support for the rare disease component of the 100,000 Genomes Project difficult to justify.

For this reason, web-based case report forms will remain available for all aspects of the data, for the full duration of the project.

Classes of Data

In the diagram below, Figure 1, the combination of sample and participant information represents the *essential sample metadata*. The remaining classes of data represent both the core data (to date) and the additional data (going forward).

This shows that each sample record is associated with a unique participant record, although there may be more than one sample record or report for a single participant: the relationship is many-to-one, indicated by the asterisk at one end of the line and the digit one at the other. Similarly, each event report is associated with a single participant, and each participant may be associated with many different events.

It shows also that the set of core data items associated with the report of an event may be extended in five different ways, depending upon the kind of event in question. If the event were a surgical intervention, for example, then a SNOMED CT or OPCS-4 code may be supplied to describe the procedure: this information would not be expected in a report of an investigation or diagnosis.

Two of these extensions or subclasses of event will be extended further, to describe the data items associated with different kinds of investigation or intervention.

The collection of extensions or subclasses of investigation and intervention, their association with specific rare diseases, and the definition of the individual data items, will be updated periodically as the project progresses.

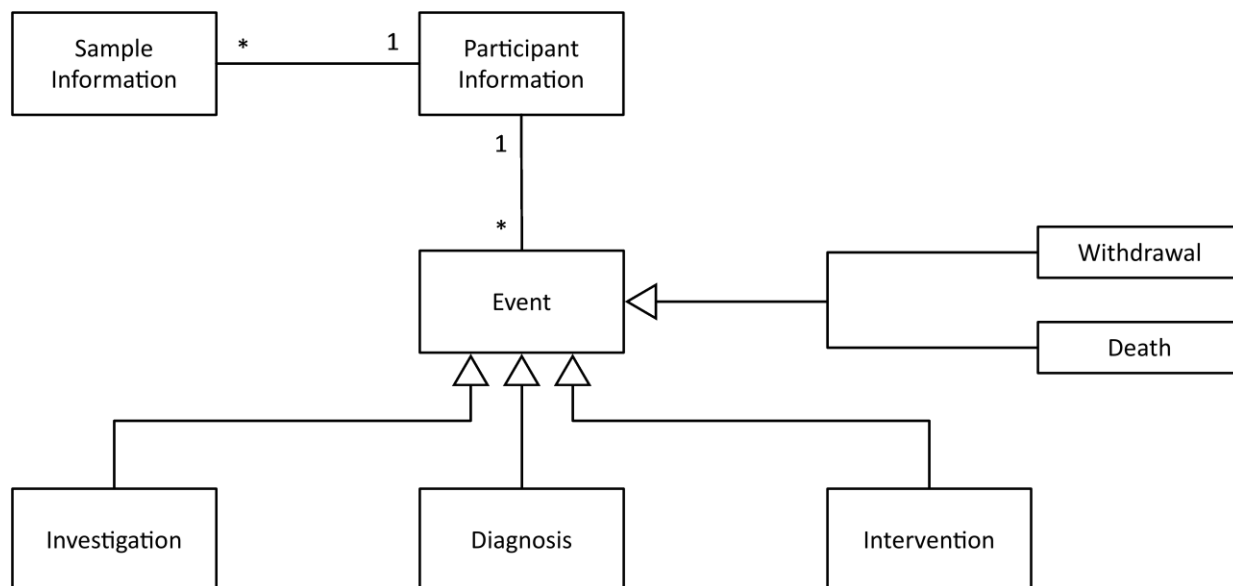


Figure 1: Classes of Rare Disease Data

Completeness

From a data modelling perspective, all of the classes are ‘optional’, in that an event of that class may not yet have occurred, or may not yet have been reported, for a particular participant.

From a contractual perspective, at least one phenotyping report, at least one pedigree diagram, at least one description of patient history, and at least one set of general observations (all subclasses of investigation) will be expected as part of the *core data*.

Beyond that, event records or reports are required for all relevant clinical events or observations to date (for the *core data*) and going forward (for the *additional data*).

For some classes of event, a report will be accepted only if values are supplied for some of the data items involved: these are the ‘mandatory’ items within those classes.

An item is ‘mandatory’ for a particular class if it has a multiplicity of 1..1 or 1..*

Where an item has a multiplicity of 0..1, a report for that class of event will be accepted even if no value has been supplied for that item.

From a contractual perspective, however, values are expected for all applicable data items, for all relevant classes of event.

File upload

Upload of image files is required for a number of data elements. These should be provided as .jpeg or .pdf format of file size <1MB unless otherwise specified.

Genomics England and NHS England are currently developing policy for collection of radiological and other non-photographic clinical image files. Details of these will be provided as soon as they are available. Such files should not be uploaded until this has been finalized.

2 Essential Sample Metadata (12499.3)

2.1 Participant Information (12500.3)

The GMC clinic is expected to establish eligibility, to validate NHS numbers before registration, to choose or identify a suitable ID as a means of referring to the family group, and to allocate an identifier as a means of referring to the participant. The data items described below, where applicable, are essential to the subsequent management of the participant, the sample, and any results obtained from the sequencing process.

2.1.1 Participant (12501.3)

Name	Description	Multiplicity	Data Type
Family ID (12504.3)	Genomics England Family Identifier assigned to the proband and their relatives. This should be the Proband Participant ID.	1..1	gmcFamilyId
Person Stated Gender (12509.1)	The participant's current gender	1..1	personStatedGenderCode <i>Enumeration</i> 2:Female 1:Male 9:Not Specified (Unable to be classified as either male or female) X:Not Known (PERSON STATED GENDER CODE not recorded)
Person Phenotypic Sex (12510.1)	The participant's phenotypic sex classification at birth	1..1	personPhenotypicSexClassification <i>Enumeration</i> 2:Female 1:Male 9:Indeterminate
Person Karyotypic Sex (31267.2)	The participant's karyotypic sex	0..1	personKaryotypicSexClassification <i>Enumeration</i> XY:XY XX:XX XO:XO XXY:XXY XYY:XYY XXX:XXX XXYY:XXYY XXXY:XXXY XXXX:XXXX

			other:other unknown:unknown
Ethnicity (14445.2)	The ethnicity of a PERSON, as specified by the PERSON. The 16+1 ethnic data categories defined in the 2001 census is the national mandatory standard for the collection and analysis of ethnicity.	1..1	ethnicCategory >10 Enumerations please click above for more details
Surname at Birth (12511.3)	The participant's surname at birth, if available and different from current surname	0..1	personFamilyName(atBirth)
Genomic Medicine Centre ID (14860.2)	ODS code of the NHS trust responsible for the patient (the main GMC trust, or the local delivery partner)	1..1	organisationCode
Local Case Identifier (33982.1)	Optional case identifier or family number used locally, if different from family id	0..1	xs:string

together with,

2.1.1.1 Participant Identifiers (29049.2)

The following information is used to identify the participant and must be included with all data submissions

Name	Description	Multiplicity	Data Type
Participant ID (12502.2)	Participant Identifier (supplied by Genomics England)	1..1	participantId
Date of Birth (12505.1)	The participant's date of birth	1..1	xs:date
NHS Number (12506.2)	Validated NHS number for participant	1..1	nhsNumber
CHI Number (14821.3)	Scottish Community Health Index (CHI) number for participant	0..1	chiNumber
Hospital Number (14506.1)	Hospital number of the participant	0..1	organisationCode
Surname (12507.2)	The participant's surname	1..1	personFamilyName
Forenames (12508.2)	The participant's forenames	1..1	personGivenName

2.1.2 Contact Details (12528.2)

Participant's contact details

Name	Description	Multiplicity	Data Type
Participant Email Address (12529.1)	Email address of participant	0..1	emailAddress
Participant Home Telephone (12532.2)	If available, the participant's home telephone number	0..1	xs:string
Participant Mobile Telephone (12533.2)	If available, the participant's mobile telephone number	0..1	xs:string

together with, in the case of

2.1.2.1 Person Address (12821.2)

Name	Description	Multiplicity	Data Type
Address line 1 (12822.1)	Premises ID and/or house name, e.g. 'Flat 1', 'The Old Schoolhouse'	0..1	addressLine
Address line 2 (12823.1)	House number, dependent thoroughfare name and descriptor without commas, e.g. '23 Mill Lane'	0..1	addressLine
Address line 3 (12824.1)	Dependent locality/village, e.g. 'Boxgrove'	0..1	addressLine
Address line 4 (12825.1)	Post town, e.g. 'Leeds'	0..1	addressLine
Address line 5 (12826.1)	County (if present), e.g. 'Hampshire', 'Hants'	0..1	addressLine
Postcode (12827.1)	The UK format Postcode, 8 character string, as per BS7666. The 8 characters field allows a space to be inserted to differentiate between the inward and outward segments of the code, enabling full use to be made of Royal Mail postcode functionality.	0..1	Postcode

2.1.3 Consent (12541.5)

A report of information obtained at consent, including the metadata associated with the report, the overall consent status (consent given) and the individual questions and responses included in the consent form (consent details)

Name	Description	Multiplicity	Data Type
Date of Consent (14535.1)	Date consent taken	1..1	xs:date
Name and Version of Consent Form (12543.2)	Name and Version of form used - list of names and versions available from genomicsengland.co.uk/library-and-resources/	1..1	genomicsEnglandConsentForms
Consent Given (12545.2)	Yes no answer to consent given	1..1	yesNo <i>Enumeration</i> yes: Yes no: No
Consent Form (12546.3)	File name of uploaded PDF copy of consent form - requested format [ParticipantId]_consent_[TimeStamp].pdf	0..1	xs:string
Person Taking Consent (12547.3)	The full name of the person taking consent	1..1	xs:string
Name and Version of Participant Information Sheet (12544.4)	Name and Version of information sheet presented - list of names and versions available from genomicsengland.co.uk/library-and-resources/	1..1	genomicsEnglandParticipantInformationSheets

together with, for each of the questions on the consent form

2.1.3.1 Consent Details (29742.1)

Specific consent details corresponding to the questions and responses on the consent form. One or more consent details will be included as part of a consent report.

Name	Description	Multiplicity	Data Type
Consent question (14546.2)	A particular question on a consent form	1..1	xs:string

Consent response (14549.3)	Yes or no response to a particular question on the consent form	1..1	yesNo <i>Enumeration</i> yes: Yes no: No
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2.1.4 Clinical Information (12553.2)

Name	Description	Multiplicity	Data Type
Participant Type (12554.2)	Type of participant - rare disease proband or participating relative	1..1	participantType <i>Enumeration</i> Proband: Proband Relative: Relative

together with, for each **proband**,

2.1.4.1 Proband (14854.2)

Name	Description	Multiplicity	Data Type
Out of Area Recruitment (35540.1)	Flag if a relative of the proband is recruited out of area	1..1	yesNo <i>Enumeration</i> yes: Yes no: No
Consanguinity (12557.2)	Presence of consanguinity	0..1	Consanguinity <i>Enumeration</i> U: Unknown P: Possible N: No Y: Yes
Pattern of affected individuals in family (31270.1)	Pattern of affected individuals in the family.	0..1	familyAffectionPattern <i>Enumeration</i> singleUnknown: Single affected person with unknown family history singleIsolated: Single isolated case of disease with unaffected parents multipleSibsNotParents: Multiple affected full siblings with unaffected parents multipleNotAllSibs: Multiple affected related individuals, not all of whom are full siblings

Penetrance (31272.2)	Is there any indication that the disease is not fully penetrant?	0..1	penetrance <i>Enumeration</i> N: No 9: Unknown Y: Yes
Group Type (12555.5)	The type of family group used against supplied enumeration	0..1	groupType <i>Enumeration</i> 1: Trio with Mother AND Father 2: Trio with Mother OR Father AND other biological relative 3: Trio with other biological relatives 4: Duo with Mother OR Father 5: Duo with other biological relative 6: Families with more than 3 participants 7: Singleton
Mother Affected (12558.3)	Mother affected with same condition as proband, chosen from supplied enumeration	1..1	affectedStatus <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Father Affected (12559.3)	Father affected with same condition as proband, chosen from supplied enumeration	1..1	affectedStatus <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Full brothers affected (12560.2)	Number of full brothers with same condition	0..1	positiveInteger
Total full brothers (12561.2)	Total number of full brothers	0..1	positiveInteger
Full sisters affected (12562.2)	Number of full sisters with same condition	0..1	positiveInteger
Total full sisters (12563.2)	Total number of full sisters	0..1	positiveInteger

and, for each **relative**

2.1.4.2 Relative (14855.2)

Name	Description	Multiplicity	Data Type
Biological Relationship to Proband (12564.4)	Biological Relationship to Proband	1..1	biologicalFamilyRelationship <i>Enumeration</i> >10 Enumerations please click above for more details
Other Biological Relationship to Proband (12565.2)	Biological Relationship to Proband if not in the enumeration supplied	0..1	xs:string
Proband Forenames (33983.1)	The forenames of the proband associated with the family group, used for consistency checks	1..1	xs:string
Proband Surname (33984.1)	The surname of the proband associated with the family group, used for consistency checks	1..1	xs:string
Proband NHS Number (33985.1)	The nhs number of the proband associated with the family group, used for consistency checks	1..1	xs:string
Proband Date of Birth (33986.1)	The date of birth of the proband associated with the family group, used for consistency checks	1..1	xs:date

2.1.5 Disease and Eligibility (12579.3)

The following disease and eligibility information must be completed for every participant

2.1.5.1 Eligibility (29284.2)

Name	Description	Multiplicity	Data Type
Eligibility Statement Version (15017.2)	Name and version of the disease specific eligibility statement - requested format [diseaseName]_[eligibilityId]	1..1	xs:string
Eligible (15018.3)	Participant's eligibility	1..1	yesNo <i>Enumeration</i> yes: Yes no: No

together with, for every affected participant,

2.1.5.2 Diagnoses (31151.3)

Diagnoses, either from the list of Genomics England rare disorders and/or Snomed CT codes and/or ICD codes. At least one GEL disorder should be included for every proband. Multiple diagnoses can be provided but each report should contain the date of the diagnosis and an event reference.

NOTE: HPO terms should be provided to supplement these diagnoses and/or where diagnosis cannot be captured within SNOMED CT / OMIM / ICD

Name	Description	Multiplicity	Data Type
Event Reference (14858.3)	Unique identifier for local record of clinical event or observation	0..1	xs:string
Event Date (12727.4)	Date of the clinical event or observation being reported e.g. date biopsy was taken	1..1	xs:dateTime

together with, for every affected participant with a disease included in the list of GEL rare disorders,

2.1.5.2.1 Disease Information (GEL) (29283.3)

Name	Description	Multiplicity	Data Type
Disease Group (12580.1)	Top-level classification of rare diseases (project specific) See Genomics England Rare Disease List	1..1	xs:string
Disease Subgroup (12581.1)	Narrower classification of disease See Genomics England Rare Disease List	1..1	xs:string
Specific Disease (12582.1)	Specific rare disorder within this classification See Genomics England Rare Disease List	1..1	xs:string
Age of Onset (12583.3)	Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders.	1..1	years(fractions)

and for every participant with a disease not in the list of GEL rare disorders,

2.1.5.2.2 Disease Information (Other) (33631.1)

Diagnoses that are not part of the Genomics England disorders

Name	Description	Multiplicity	Data Type
Medical Condition (ICD10) (31155.1)	Medical condition coded using ICD10	0..1	ICD10Code
Medical Condition (SnomedCT) (31153.1)	Medical condition coded using SnomedCT	0..1	diagnosticTermsSnomedCT
OMIM Code (29827.1)	OMIM code that best describes the disorder	0..1	xs:string

together with, for each **proband**,

2.1.5.3 Consultant Details (14515.3)

Details of the consultant responsible for the patients care

Name	Description	Multiplicity	Data Type
Full Name of Responsible Consultant (12774.4)	Nominated person responsible for patients clinical care and recipient of clinical reports and communications for Genomics England	1..1	xs:string
Consultant GMC number (31254.1)	GMC number of consultant with responsibility for the patient's clinical care	0..1	xs:string
Full Name not Consultant (14517.3)	Full name of person entering data on behalf of consultant	0..1	xs:string
Contact number (14520.3)	Phone number for the consultant.	0..1	xs:string
Hospital of Responsible Consultant (12516.2)	ODS code of the hospital to which the consultant is contracted under their MAIN SPECIALTY for the purposes of the current work.	0..1	organisationCode

2.1.6 Family (12586.3)

For all participants (for findings and feedback),

Name	Description	Multiplicity	Data Type
Mothers Ethnic Origin (12587.2)	The ethnic origin of the participant's father, against supplied enumeration. If no origin supplied please use Z	1..1	ethnicCategory <i>Enumeration</i>

Mothers Ethnic Origin Other (12588.2)	Ethnic origin, if not in enumeration supplied	0..1	xs:string
Mothers Other Relevant Ancestry (12589.2)	Pertinent to clinical findings, additional relevant ancestry such as ashkenazi	0..1	xs:string
Fathers Ethnic Origin (12595.2)	The ethnic origin of the participant's father, against supplied enumeration. If no origin supplied please use Z	1..1	ethnicCategory <i>Enumeration</i> >10 Enumerations please click above for more details
Fathers Ethnic Origin Other (12596.4)	Father's ethnic origin, if not in enumeration supplied	0..1	xs:string
Fathers Other Relevant Ancestry (12597.2)	Pertinent to clinical findings, additional relevant ancestry such as ashkenazi	0..1	xs:string
Maternal Family History of Breast and/or Ovarian Cancer (12590.3)	If available, maternal first or second degree relative with history of Breast and/or Ovarian Cancer, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Maternal Family History of Colorectal Cancer (12591.3)	If available, maternal first or second degree relative with history of Colorectal Cancer, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Maternal Family History of Ischaemic Heart Disease or Stroke (12592.3)	If available, maternal first or second degree relative with history of Ischaemic Heart Disease or Stroke, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Maternal Family History of Endocrine Tumours (12593.3)	If available, maternal first or second degree relative with history of Endocrine Tumours, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Other Relevant Maternal Family History (12594.1)	Other relevant history	0..1	xs:string
Paternal Family History of Breast	If available, paternal first or second degree relative with history of	1..1	yesNoUnk <i>Enumeration</i>

and/or Ovarian Cancer (12598.3)	Breast and/or Ovarian Cancer if unknown please choose unknown.		yes: Yes no: No unknown: Unknown
Paternal Family History of Colorectal Cancer (12599.3)	If available, paternal first or second degree relative with history of Colorectal Cancer, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Paternal Family History of Ischaemic Heart Disease or Stroke (12600.3)	If available, paternal first or second degree relative with history of Ischaemic Heart Disease or Stroke, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Paternal Family History of Endocrine Tumours (12601.3)	If available, paternal first or second degree relative with history of Endocrine Tumours, if unknown please choose unknown.	1..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Other Relevant Paternal Family History (12602.1)	Other Relevant Paternal Family History	0..1	xs:string

2.2 Sample Information (29287.3)

For each blood sample successfully processed, the GMC laboratory will provide a set of sample metadata to Genomics England, at or before the point when the derived products are dispatched to the GEL Biorepository

Name	Description	Multiplicity	Data Type
Participant ID (12502.2)	Participant Identifier (supplied by Genomics England)	1..1	participantId
Sample ID (12607.3)	The barcode from the sample tube used for sample collection at the GMC clinic. This identifier should be unique within a GMC clinic.	1..1	sampleId
Clinic ID (12503.1)	ODS code for GMC clinic	1..1	organisationCode
Clinic Sample Type (12616.3)	The type of the sample (against specified enumeration)	1..1	gmcSampleType <i>Enumeration</i> >10 Enumerations please click above for more details
Clinic Sample DateTime (12617.1)	Timestamp containing the date and time the sample was taken in clinic. This must be captured on the sample linkage form and transferred into the GMC LIMS.	1..1	xs:dateTime
Laboratory ID (12619.1)	ODS code of the laboratory organisation used for sample processing i.e. extraction, QC, collection or dispatch N.B. this could relate to a Blood Extraction Facility for QC data, otherwise we expect this to be a GMC Laboratory	1..1	organisationCode
Laboratory Sample ID (12621.3)	(if dispatched) the 2D barcode from the laboratory sample tube used for dispatch to the GEL Biorepository. This identifier should be unique within a GMC Lab.	1..1	laboratorySampleId
Laboratory Method (12623.2)	Version of Genomics England protocol used for sample handling and processing	1..1	gelSampleProtocolVersion <i>Enumeration</i> v1:v1 of the Genomics England sample handling

			and processing protocol v2:v2 of the Genomics England sample handling and processing protocol
GMC Rack Well (12893.1)	The GMC must record the position of the sample in the Rack they send to the biorepository. Each rack is has 96 wells. The position of a sample in these wells is coded from from A-H on the x-axis (short side) and 1-12 on the y-axis (long side) i.e. A3	1..1	rackWell
GMC Sample Dispatch Date (12626.3)	(if dispatched) the date upon which the sample is dispatched to the GEL Biorepository	1..1	xs:dateTime
GMC Sample Consignment Number (12627.1)	(if dispatched) the consignment number used by the transport service	1..1	xs:string
GMC Rack ID (12625.2)	(if dispatched) the barcode on the containing rack as dispatched	1..1	rackId

2.2.1 Test Results (29288.2)

For each sample successfully processed, the GMC laboratory will provide a set of test results to Genomics England, at or before the point when the derived products (in particular, the extracted DNA) are dispatched to the GEL Biorepository.

Name	Description	Multiplicity	Data Type
Test Result DateTime (12609.1)	The date and time that the test results were obtained	1..1	xs:dateTime
Test Result Type (12608.5)	QC test result type	1..1	gmcTestResults <i>Enumeration</i> >10 Enumerations please click above for more details
Test Result Value (12610.1)	The value obtained	1..1	xs:string
Test Result Source (31144.1)	Source of the test result - GMC, Biorepository or Sequencer	1..1	sourceOrganisation <i>Enumeration</i> GMC: Genomic Medicine Centre BIO: Biorepository

			SEQ:Sequencer
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2.2.2 Blood Sample (29089.3)

In addition to the sample metadata, for each blood sample successfully processed, the GMC laboratory will provide additional items of sample metadata.

Name	Description	Multiplicity	Data Type
Laboratory Sample Volume (12622.1)	(if dispatched) the volume of the product in the laboratory sample tube as dispatched	1..1	volumeInMicroliters
Laboratory Remaining Volume Banked (12624.1)	(if dispatched) the amount of additional product remaining at the laboratory (which may be zero) N.B. DNA concentration must be > 30ng/ul for normal germline samples and >10ng/ul for FF samples	1..1	volumeInMicroliters

3 Core Data (12736.3)

3.1 Participant Identifiers

The following information is used to identify the participant and must be provided with every data submission

Name	Description	Multiplicity	Data Type
Participant ID (12502.2)	Participant Identifier (supplied by Genomics England)	1..1	participantId
Date of Birth (12505.1)	The participant's date of birth	1..1	xs:date
NHS Number (12506.2)	Validated NHS number for participant	1..1	nhsNumber
CHI Number (14821.3)	Scottish Community Health Index (CHI) number for participant	0..1	chiNumber
Hospital Number (14506.1)	Hospital number of the participant	0..1	organisationCode
Surname (12507.2)	The participant's surname	1..1	personFamilyName
Forenames (12508.2)	The participant's forenames	1..1	personGivenName

3.2 Pedigree (12772.3)

Name	Description	Multiplicity	Data Type
Pedigree Diagram (12775.1)	Filename for the uploaded pedigree diagram - requested format is [ParticipantId]_pedigree_[TimeStamp]]	0..1	xs:string
Family ID (12504.3)	Genomics England Family Identifier assigned to the proband and their relatives. This should be the Proband Participant ID.	1..1	gmcFamilyId

together with, for each family member,

3.2.1 Pedigree Member (33574.2)

Name	Description	Multiplicity	Data Type
Participant ID (12502.2)	Participant Identifier (supplied by Genomics England)	0..1	participantId
Member ID (35473.1)	Internal Id used to identify family relationships between pedigree members i.e. father id / mother id	1..1	xs:integer
Father ID (35472.1)	Pedigree Id of the father, if unknown then no parent is referenced. Parents may need to be entered even if no data is known about them in order to unambiguously reconstruct the pedigree.	0..1	xs:integer
Mother ID (35471.1)	Pedigree Id of the mother, if unknown then no parent is referenced. Parents may need to be entered even if no data is known about them in order to unambiguously reconstruct the pedigree.	0..1	xs:integer
Person Phenotypic Sex (12510.1)	The participant's phenotypic sex classification at birth	1..1	personPhenotypicSexClassification <i>Enumeration</i> 2: Female 1: Male 9: Indeterminate
Forenames (12508.2)	The participant's forenames	0..1	personGivenName

Surname (12507.2)	The participant's surname	0..1	personFamilyName
NHS Number (12506.2)	Validated NHS number for participant	0..1	nhsNumber
Date of Birth (12505.1)	The participant's date of birth	0..1	xs:date
Consanguinity (14564.1)	This indicates that the participant is the product of a consanguinous relationship	1..1	Consanguinity <i>Enumeration</i> U :Unknown P :Possible N :No Y :Yes
Twin Group (35470.1)	Each twin group is numbered, i.e. all members of a group of multiparous births receive the same number	0..1	xs:integer
Monozygotic (35469.1)	If twin, are they monozygotic	0..1	yesNoUnk <i>Enumeration</i> yes :Yes no :No unknown :Unknown
Adopted Status (35468.1)	Default is not_adopted. Adopted in means adopted into the family Adoptedout means child belonged to the family and was adopted out	1..1	adoptedStatus <i>Enumeration</i> not_adopted :not adopted adopted_in :adopted into the family adopted_out :child belonged to the family and was adopted out
Life Status (35465.1)	Life Status	1..1	lifeStatus <i>Enumeration</i> alive :alive aborted :aborted deceased :deceased unborn :unborn stillborn :stillborn miscarriage :miscarriage

together with, for each phenotyping event associated with the family member,

3.2.1.1 Phenotype Report (12950.3)

See phenotype report (12950.3)

together with, in the case of for each diagnosis associated with the family member,

3.2.1.2 Diagnoses

See diagnosis (31151.3)

3.3 Investigation (29497.5)

Report of an investigation relevant to a rare disease participant. Please note each data element within a clinical investigation may have the following attributes:

- a data type;
- a set of enumerated values;
- a set of applicable measurement units;
- a normal range.

Data elements within each report may correspond to metadata about the investigation or may be values collected as part of the investigation.

All investigation reports must include the following data elements.

Name	Description	Multiplicity	Data Type
Event Date (12727.4)	Date of the clinical event or observation being reported e.g. date biopsy was taken	1..1	xs:dateTime
Event Reference (14858.3)	Unique identifier for local record of clinical event or observation	0..1	xs:string

3.3.1 Phenotype Report (12950.3)

Where applicable, indicate relevant phenotyping terms using the following report

Name	Description	Multiplicity	Data Type
Full Name of Responsible Consultant (12774.4)	Nominated person responsible for patients clinical care and recipient of clinical reports and communications for Genomics England	0..1	xs:string
Hospital of Responsible Consultant (12516.2)	ODS code of the hospital to which the consultant is contracted under their MAIN SPECIALTY for the purposes of the current work.	0..1	organisationCode
Full Name not Consultant (14517.3)	Full name of person entering data on behalf of consultant	0..1	xs:string
Clinical Assessment Letter (30853.1)	File name of uploaded copy of the most informative letter(s) or assessment relating to the patients phenotype - requested format [ParticipantId]_assessment_[TimeStamp]	0..unbounded	xs:string
Phenotype Report Code (33567.1)	Only applicable for XML submissions. This will be the identifier and version of the set of HPO terms suggested for a particular disorder i.e. for Familial Thoracic Aortic Aneurysm Disease the code would be 11021.4. If this is not recorded against a set of suggested phenotypes leave blank.	0..1	xs:string

together with, in the case of one or more **phenotyping statements**

3.3.1.1 Phenotype Statement (14845.1)

Phenotype statements that relate to the individual can be included in every phenotype report. Each of these statements must include the following elements.

Name	Description	Multiplicity	Data Type
HPO Build Number (14643.1)	This is the build number of the HPO ontology used.	0..1	xs:string
Phenotype Identifier	The identity of the statement	1..1	hpoPhenotypes

(14642.2)	within the Human Phenotype Ontology		
Phenotype Present (14646.1)	Presence of the phenotypic abnormality.	1..1	Present <i>Enumeration</i> unknown: Unknown yes: Yes no: No

together with the following modifiers, if applicable

3.3.1.2 Modifiers (12957.1)

Name	Description	Multiplicity	Data Type
Laterality (11156.1)	The localization with respect to the side of the body of the specified phenotypic abnormality.	0..1	Laterality <i>Enumeration</i> Right: Being located on the right side of the body. Unilateral: Being present on only the left or only the right side of the body. Bilateral: Being present on both sides of the body. Left: Being located on the left side of the body.
Progression (11159.1)	The progression of the phenotype, can be progressive or non-progressive	0..1	Progression <i>Enumeration</i> Progressive: Progressive Nonprogressive: Nonprogressive
Onset (11157.2)	The age group in which disease manifestations appear. Comment: Adolescent is defined by WHO as a person between 10-19 years of age.	0..1	Onset <i>Enumeration</i> Embryonal onset: Onset of disease at up to 8 weeks of gestation. Fetal onset: Onset prior to birth but after 8 weeks of gestation. Neonatal onset: Onset of signs or symptoms of disease within the first 28 days of life. Infantile onset: Onset of signs or symptoms of disease between 28 days to one year of life. Comment: Onset of signs or

			<p>symptoms of disease within the first 24 months of life.</p> <p>Childhood onset:Onset of disease at the age of between 1 and 5 years.</p> <p>Juvenile onset:Onset of signs or symptoms of disease between the age of 5 and 15 years.</p> <p>Young adult onset:Onset of disease at the age of between 16 and 40 years.</p> <p>Late Onset:A type of adult onset with onset of symptoms after the age of 60 years.</p> <p>Middle age onset:A type of adult onset with onset of symptoms at the age of 40 to 60 years. Comment: Middle age is of course not amenable to precise definitions. We suggest using this term for onset of disease symptoms between the age of 40 and 60 years.</p>
Spatial pattern (11158.2)	The pattern by which a phenotype affects one or more regions of the body.	0..1	<p>spatialPattern <i>Enumeration</i></p> <p>Distal:Localized away from the central point of the body.</p> <p>Generalized:Affecting all regions without specificity of distribution.</p> <p>Localized:Being confined or restricted to a particular location.</p> <p>Proximal:The pattern by which a phenotype affects one or more regions of the body.</p>
Severity (11155.1)	The intensity or degree of a manifestation.	0..1	<p>Severity <i>Enumeration</i></p> <p>Borderline:Having a minor degree of severity that is considered to be on the boundary between the normal and the abnormal ranges. For quantitative traits, a deviation of that is less than two standard deviations from the appropriate population mean.</p> <p>Mild:Having a relatively minor</p>

			<p>degree of severity. For quantitative traits, a deviation of between two and three standard deviations from the appropriate population mean.</p> <p>Moderate: Having a medium degree of severity. For quantitative traits, a deviation of between three and four standard deviations from the appropriate population mean.</p> <p>Severe: Having a high degree of severity. For quantitative traits, a deviation of between four and five standard deviations from the appropriate population mean.</p> <p>Profound: Having an extremely high degree of severity. For quantitative traits, a deviation of more than five standard deviations from the appropriate population mean.</p>
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3.3.2 General Observations (29290.3)

3.3.2.1 Growth Parameters (14808.4)

3.3.2.1.1 Height Details (32935.2)

Name	Description	Multiplicity	Data Type
Height (14755.3)	Person height / length in metres to 2 decimal places. Height and weight to be used to calculate BMI as an indicator of the patient being overweight or obese. Provide the most relevant information that will inform this.	1..1	personHeightInMetres
Date of measurement (14756.1)	When was the measurement taken	0..1	xs:date
Type of measurement (14759.1)	How was the measurement taken i.e. patient reported	0..1	typeOfMeasurement <i>Enumeration</i> PatientReported :Patient reported Measured :Measured Unknown :Unknown

3.3.2.1.2 Weight Details (32943.2)

Name	Description	Multiplicity	Data Type
Weight (14760.2)	Weight in kg. Height and weight to be used to calculate BMI as an indicator of the patient being overweight or obese. Provide the most relevant information that will inform this.	1..1	personObservation(weight)
Date of measurement (14756.1)	When was the measurement taken	0..1	xs:date
Type of measurement (14759.1)	How was the measurement taken i.e. patient reported	0..1	typeOfMeasurement <i>Enumeration</i> PatientReported :Patient reported Measured :Measured Unknown :Unknown

3.3.2.1.3 OFC Details (32945.1)

Name	Description	Multiplicity	Data Type
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OFC (14761.2)	Head circumference or occipitofrontal circumference in centimetres	1..1	OFC
Date of measurement (14756.1)	When was the measurement taken	0..1	xs:date
Type of measurement (14759.1)	How was the measurement taken i.e. patient reported	0..1	typeOfMeasurement <i>Enumeration</i> PatientReported :Patient reported Measured :Measured Unknown :Unknown

3.3.2.2 Age of onset under 16 (14765.4)

The following information should be provided for each individual with a disorder with onset under the age of 16.

3.3.2.2.1 Pregnancy details (14766.4)

Name	Description	Multiplicity	Data Type
Parents with at least 3 miscarriages (14769.3)	Did the parents of the proband have three or more miscarriages?	0..1	yesNoUnk <i>Enumeration</i> yes :Yes no :No unknown :Unknown
Conception (14772.1)	Type of conception	0..1	Conception <i>Enumeration</i> Spontaneous :Spontaneous Other :Other InVitro :In vitro fertilisation
Number of fetuses in the pregnancy (14773.1)	Number of fetuses in the pregnancy	0..1	positiveInteger

3.3.2.2.2 Birth details (14782.3)

Name	Description	Multiplicity	Data Type
Gestation (14783.2)	The gestation length of a Fetus Episode recorded as the total number of weeks. The calculation may be: a) calculated by ultrasound scan measurements according to	0..1	xs:integer

	<p>the trimester of the scan</p> <p>b) estimated from the LAST MENSTRUAL PERIOD DATE</p> <p>c) estimated by clinical assessment (in the absence of a or b)</p> <p>The number of completed whole weeks of gestation.</p>		
Weight in Kilogrammes at birth (14786.2)	Weight at Birth in kilogrammes	0..1	personObservation(weight)
Birth OFC (14789.1)	Head circumference or occipitofrontal circumference at birth	0..1	OFC
Admission to special care (14792.2)	Duration of admission to special care baby unit or neonatal intensive care unit in weeks	0..1	Weeks

3.3.2.2.3 Developmental milestones (14795.2)

Name	Description	Multiplicity	Data Type
Social smile (14796.1)	Length of time in months between birth and first social smile	0..1	Months
Sat independently (14799.1)	Length of time in months between birth and sitting independently	0..1	Months
Walked independently (14802.1)	Length of time in months between birth and walking independently	0..1	Months
First words (14805.2)	Length of time in months between birth and first words spoken with meaning	0..1	Months

3.3.3 Genetic Investigations (12759.3)

In addition to the core essential investigation data items, genetic investigations relating to germline molecular genetics should have the following:

Name	Description	Multiplicity	Data Type
Sample Taken Date (12760.2)	The date upon which the sample was taken	0..1	xs:dateTime
Source Sample ID (12762.1)	The local identifier for the source sample	0..1	localSampleId
Sample Receipt Date (12761.2)	The date upon which the sample was received at the laboratory	0..1	xs:dateTime
Sample tissue of origin (33274.1)	Origin of the sample tissue	1..1	tissueOrigin <i>Enumeration</i> Blood: Blood Skin: Skin Saliva: Saliva Muscle: Muscle CVS: CVS Amniocentesis: Amniocentesis Tumour: Tumour Other: Other
Assessment (29524.1)	Assessment of findings and clinical significance	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

3.3.3.1 Genetic Results (14856.4)

If the investigation produced genetic results, then the genetic investigation should be extended with the following items, for each genetic result

GUIDANCE: Enter all abnormal genetic results and all pertinent negative results from this sample. Use one entry per gene.

3.3.3.1.1 Genetic Result (31148.3)

Name	Description	Multiplicity	Data Type
Genetic Test Laboratory (15020.3)	Was this test performed in a diagnostic or research laboratory?	0..1	geneticTestLaboratory <i>Enumeration</i> research_laboratory :Research laboratory diagnostic_laboratory :Diagnostic laboratory
Test Scope (12763.3)	The gene coded according to HGNC. Enter 'genomewide' if genomewide, e.g. karyotype or aCGH.	1..1	geneScope
Scope Qualifiers (12764.2)	If whole locus or coding sequence of gene not covered, give details of regions covered, e.g. 'exons 3 and 8'	0..1	xs:string
Method of Test (12765.3)	The method used to investigate the gene(s). If copy number analysis has been performed for a subset of genes, please enter separately from sequencing results	1..1	geneticTestMethod <i>Enumeration</i> >10 Enumerations please click above for more details
Test Result (12744.2)	(for molecular results) If no defect was observed please report 'normal'; if a mutation is detected that is considered pathogenically or clinically important record 'mutation detected'; if no reliable result could be determined please report 'fail'.	1..1	geneticTestResult <i>Enumeration</i> Normal :Normal (negative) Fail :Fail AbnormalityDetected :Pathogenic abnormality detected VUS :Variant of unknown significance detected
Details of Abnormal Result (14900.3)	Record the details of the abnormal genotype using Genomic Coordinates	0..1	xs:string
Genome Build for abnormal molecular result (34224.2)	Record the relevant human genome build if an abnormal genotype is specified if applicable	0..1	xs:string
Details of Abnormal Cytogenetic Result (34225.1)	Record the details of the cytogenetic abnormality using IGCN standards	0..1	xs:string

3.3.3.2 Genetic Reports (15019.5)

if a genetic report was produced, then the initial list of five items should be extended with

GUIDANCE: Upload all genetic test reports from this sample

Name	Description	Multiplicity	Data Type
Genetic Test Laboratory (15020.3)	Was this test performed in a diagnostic or research laboratory?	0..1	geneticTestLaboratory <i>Enumeration</i> research_laboratory :Research laboratory diagnostic_laboratory :Diagnostic laboratory
Description of Genetic Test (15153.1)	Please describe the genetic test performed. If available, please use the name provided by the UK Genetic Testing Network http://ukgtn.nhs.uk/	0..1	xs:string
Test Result (12744.2)	(for molecular results) If no defect was observed please report 'normal'; if a mutation is detected that is considered pathogenically or clinically important record 'mutation detected'; if no reliable result could be determined please report 'fail'.	1..1	geneticTestResult <i>Enumeration</i> Normal :Normal (negative) Fail :Fail AbnormalityDetected :Pathogenic abnormality detected VUS :Variant of unknown significance detected
Genetic Report (15027.4)	File name of uploaded copy of report - requested format [ParticipantId]_molecular_report_[TimeStamp]	1..unbounded	xs:string
Sequence File (12767.2)	Local sequence file reference or uploaded copy of VCF	0..1	xs:string

3.3.4 Biopsy (29498.2)

All biopsy reports should include the following elements,

Name	Description	Multiplicity	Data Type
Biopsy Procedure Other (snomedCT) (29735.1)	Biopsy procedure, if not included in current list (SnomedCT)	0..1	procedureCodeSnomedCT
Biopsy Procedure Other (OPCS-4) (30851.1)	Biopsy procedure if not included in current list (OPCS-4)	0..1	OPCS-4
Analysis Type (29500.2)	Type of analysis performed on the biopsy	0..1	biopsyAnalysisType <i>Enumeration</i> Light Microscopy: Light Microscopy Immunohistochemistry: Immunohistochemistry Electron Microscopy: Electron Microscopy Biochemistry: Biochemistry Respiratory chain enzyme analysis: Respiratory chain enzyme analysis
Report (29501.2)	File name of uploaded copy of clinical report - requested format [Participant ID]_[Local Report Identifier]	0..1	xs:base64Binary
Assessment (29524.1)	Assessment of findings and clinical significance	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Biopsy Findings (Pathology) (29642.1)	Snomed ct codes for overall conclusion of biopsy	0..unbounded	xs:string
Patient Status (34004.1)	Represents the patient's status when test was performed	1..1	patientStatus <i>Enumeration</i> presenting: Taken at patient presentation diagnostic: Taken at the point of diagnosis baseline: Representing a baseline measurement most abnormal: Representing the most

			abnormal measurement unknown: Status unknown
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together with, in the case of

3.3.4.1 General Biopsy (33614.1)

General investigation report to capture biopsies

Name	Description	Multiplicity	Data Type	Same As
Biopsy Procedure (29499.2)	Procedure employed for biopsy, chosen from list	1..1	biopsyProcedure <i>Enumeration</i> Biopsy of peripheral nerve: Biopsy of peripheral nerve Biopsy of peritoneum: Biopsy of peritoneum Biopsy of skin and/or subcutaneous tissue: Biopsy of skin and/or subcutaneous tissue Biopsy of skin for fibroblast culture: Biopsy of skin for fibroblast culture Biopsy or scrape of nasal cilia: Biopsy or scrape of nasal cilia Biopsy of skeletal muscle: Biopsy of skeletal muscle Bone marrow biopsy, needle or trocar: Bone marrow biopsy, needle or trocar Kidney biopsy: Kidney biopsy Transplant kidney biopsy: Transplant kidney biopsy Other: Other	

or, in the case of

3.3.4.2 Renal Biopsy (30153.2)

Name	Description	Multiplicity	Data Type
Biopsy Procedure (33617.1)	Renal Biopsy Procedure employed for biopsy, chosen from list	1..1	Renal Biopsy Procedure <i>Enumeration</i> Kidney biopsy: Kidney biopsy Other: Other
C1q immunostain result (30165.1)	Result of a C1q immunostain test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
C3 immunostain result (30166.1)	Result of a C3 immunostain test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Congo red result (30167.1)	Result of a Congo red test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Electron dense deposits result (30168.1)	Electron dense deposit result	0..1	electronDenseDeposits <i>Enumeration</i> mesangial: Mesangial subendothelial: Subendothelial subepithelial: Subepithelial intramembranous: Intramembranous
Glomerular basement membrane morphology result (30169.1)	Glomerular basement membrane morphology result	0..1	basementMembrane <i>Enumeration</i> normal: Normal thinned: Thinned abnormal: Abnormal
IgA immunostain result (30170.1)	Result of a IgA immunostain test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown

IgG immunostain result (30171.1)	Result of a IgG immunostain test	0..1	yesNoUnk <i>Enumeration</i> yes :yes no :no unknown :unknown
IgM immunostain result (30172.1)	Result of a IgM immunostain test	0..1	yesNoUnk <i>Enumeration</i> yes :yes no :no unknown :unknown
Podocyte foot process effacement result (30173.1)	Podocyte foot process effacement result	0..1	yesNoUnk <i>Enumeration</i> yes :yes no :no unknown :unknown
Tubular basement membrane morphology result (30174.1)	Tubular basement membrane morphology result	0..1	basementMembrane <i>Enumeration</i> normal :Normal thinned :Thinned abnormal :Abnormal
Tubulointerstitial infiltrate result (30175.1)	Tubulointerstitial infiltrate result (free text - will be updated in due course)	0..1	xs:string
Protein Deposition Result (30849.1)	Result for protein deposition in renal biopsy	0..1	proteinDeposition <i>Enumeration</i> amyloid :Amyloid fibrin :Fibrin immunotactoid :Immunotactoid other :Other

or, in the case of

3.3.4.3 Nasal Cilia Imaging (29830.2)

Name	Description	Multiplicity	Data Type
Biopsy Procedure (33624.1)	Nasal Cilia Procedure employed for biopsy, chosen from list	1..1	Nasal Cilia Procedure <i>Enumeration</i>

			Biopsy or scrape of nasal cilia: Biopsy or scrape of nasal cilia Other: Other
Ciliary beat frequency (28763.1)	Ciliary beat frequency in Hz	0..1	beatFrequencyInHz
Other ciliary anomaly (31284.1)	Other ciliary anomaly	0..unbounded	xs:string

3.3.5 Imaging Diagnostics (29736.2)

All imaging diagnostics reports should include the following elements,

Name	Description	Multiplicity	Data Type
Imaging Modality Other (SNOMED CT) (29738.1)	Imaging modality if not in current list (as SNOMED CT)	0..1	procedureCodeSnomedCT
Imaging Modality Other (OPCS-4) (30855.1)	Imaging modality if not in current list (as OPCS-4)	0..1	OPCS-4
Sub Region (29744.1)	Subregion of body imaged (as SNOMED CT)	0..1	bodyStructure
Assessment (29524.1)	Assessment of findings and clinical significance	1..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Fetal Classification (SNOMED CT) (29868.1)	Descriptive classification of imaging on fetal body structures, based on SNOMED CT codes	0..1	fetalBodyStructuresSnomedCt
Fetal Classification (Other) (30850.1)	Descriptive classification of imaging on fetal body structures, as free text or in code set other than SNOMED CT	0..1	xs:string
Report (29501.2)	File name of uploaded copy of clinical report - requested format [Participant ID]_[Local Report Identifier]	0..1	xs:base64Binary
Image File (12755.2)	File name of uploaded copy of image – requested format [ParticipantID]_[Local Report Identifier]	0..1	xs:base64Binary
Patient Status (34004.1)	Represents the patient's status when test was performed	1..1	patientStatus <i>Enumeration</i> presenting: Taken at patient presentation

			diagnostic: Taken at the point of diagnosis baseline: Representing a baseline measurement most abnormal: Representing the most abnormal measurement unknown: Status unknown
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together with, in the case of

3.3.5.1 General Imaging Diagnostics (33633.1)

General imaging diagnostics report to capture reports that aren't included in the specific subclasses.

Name	Description	Multiplicity	Data Type
Imaging Modality (29737.2)	Imaging modality, chosen from a list	1..1	imagingProcedure Enumeration >10 Enumerations please click above for more details
Imaging Submodality (29861.2)	Indicate the sub-modality of imaging technique used	0..1	imagingSubModality Enumeration >10 Enumerations please click above for more details
Region (29864.2)	Region of body imaged, chosen from list	1..1	bodyRegion Enumeration >10 Enumerations please click above for more details
System (29866.1)	Category of system investigated by imaging	0..1	bodySystem Enumeration >10 Enumerations please click above for more details

or, in the case of

3.3.5.2 Echocardiogram (29800.2)

Applicable to all echocardiograms, i.e. cardiomyopathy and CTD.

Name	Description	Multiplicity	Data Type
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Imaging Modality (33636.1)	Imaging Modality - Echocardiogram	1..1	Imaging Modality (Echocardiogram) <i>Enumeration</i> Diagnostic Ultrasonography: Diagnostic Ultrasonography Other: Other
Imaging Submodality (33639.1)	Imaging Submodality Echocardiogram	1..1	Imaging Submodality (Echocardiogram) <i>Enumeration</i> Echocardiogram: Echocardiogram Other: Other
Region (33642.1)	Region (Echocardiogram)	1..1	Region (Echocardiogram) <i>Enumeration</i> Heart: Heart
System (33645.1)	System Echocardiogram	1..1	System (Echocardiogram) <i>Enumeration</i> Cardiovascular: Cardiovascular
LVEDD (15223.1)	State the LEFT VENTRICULAR DIASTOLIC DIAMETER measured during the ECHOCARDIOGRAM in 2D mode	0..1	xs:decimal
LVESD (15225.1)	State the LEFT VENTRICULAR SYSTOLIC DIAMETER measured during the ECHOCARDIOGRAM in 2D mode	0..1	xs:decimal
LV EJECTION FRACTION (15221.1)	State the EJECTION FRACTION of the LEFT VENTRICLE as a percentage	0..1	xs:decimal
LVEDV (LV end diastolic volume) (27773.1)	State the LEFT VENTRICULAR END DIASTOLIC VOLUME in ml	0..1	volumeInMl
LVESV (LV end systolic volume) (27774.1)	State the LEFT VENTRICULAR END SYSTOLIC VOLUME in ml	0..1	volumeInMl
Maximum LV thickness (27778.1)	The maximum thickness of the LEFT VENTRICLE in mm	0..1	thicknessInMm
LV septal thickness diastole (28516.1)	Thickness of LEFT VENTRICULAR SEPTUM during DIASTOLE in mm	0..1	thicknessInMm

LV posterior wall thickness diastole (28517.1)	Thickness of LEFT VENTRICULAR POSTERIOR WALL during DIASTOLE in mm	0..1	thicknessInMm
Left atrium diameter (28520.1)	Diameter of LEFT ATRIUM in mm	0..1	diameterInMm
Left atrial area (28523.1)	Area of LEFT ATRIUM in cm2	0..1	areaInCm2
Hypertrophy exclusively at the apex (29801.1)	Hypertrophy exclusively at the apex present	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Pattern of LV hypertrophy (28529.2)	Pattern of LEFT VENTRICULAR hypertrophy	0..1	patternOfLvHypertrophy <i>Enumeration</i> Symmetric: Symmetric Asymmetric (maximum wall thick/min wall thick of >1.3): Asymmetric (maximum wall thick/min wall thick of >1.3) Unknown: Unknown
LV outflow tract gradient at rest (28533.1)	LEFT VENTRICULAR outflow tract gradient at rest in mmHg	0..1	pressureInMmhg
LV outflow tract gradient with valsalva (28534.1)	LEFT VENTRICULAR outflow tract gradient with valsalva in mmHg	0..1	pressureInMmhg
RV dilation (28535.2)	Is there any dilation of the RIGHT VENTRICLE?	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
RV global systolic dysfunction (28536.2)	Is global systolic RIGHT VENTRICULAR dysfunction present?	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
RV hypertrophy (28537.2)	Is there evidence of RIGHT VENTRICULAR hypertrophy?	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown

RV Aneurysms (29802.1)	Right Ventricular Aneurysms	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Maximum RV wall thickness (28538.1)	Maximum RIGHT VENTRICULAR wall thickness in mm	0..1	thicknessInMm
Bicuspid valve disease (27771.2)	Does the echocardiogram show evidence of bicuspid valve disease?	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Pulmonary valve regurgitation (31475.1)	Is pulmonary valve regurgitation present on investigation	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Aortic root diameter at sinuses of Valsalva (28561.1)	Diameter of aortic root during Valsava in mm	0..1	diameterInMm
Maximum thoracic aortic diameter (28562.1)	Maximum thoracic aorta diameter in mm	0..1	diameterInMm
Cardiac structural disease found (31471.1)	Structural disease found on cardiac investigation	0..1	yesnunknown <i>Enumeration</i> yes: yes no: no unknown: unknown
Cardiac structural disease description (31472.1)	Description of structural disease found in cardiac investigation	0..1	xs:string

or, in the case of

3.3.5.3 Kidney Imaging (29807.2)

Name	Description	Multiplicity	Data Type
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Imaging Modality (34290.1)	Imaging Modality for Renal imaging	1..1	Imaging Modality (Kidney Imaging) <i>Enumeration</i> Computerised Axial Tomography: Computerised Axial Tomography Diagnostic Ultrasonography: Diagnostic Ultrasonography Magnetic Resonance Imaging: Magnetic Resonance Imaging Nuclear Medicine: Nuclear Medicine Plain Radiography: Plain Radiography Positron Emission Tomography: Positron Emission Tomography Single Photon Emission Computed Tomography: Single Photon Emission Computed Tomography SAP scan: SAP scan Other: Other
Imaging Submodality (34291.1)	Imaging Submodality for renal imaging	0..1	Imaging Submodality (Kidney Imaging) <i>Enumeration</i> DEXA: dual energy X-ray absorptiometry DMSA: dimercaptosuccinic acid scan Micturating cystourethrogram: Micturating cystourethrogram
Region (33734.1)	Kidney Region	1..1	kidneyRegion <i>Enumeration</i> Abdominal: Abdominal
System (33737.1)	Kidney System	1..1	kidneySystem <i>Enumeration</i> Genitourinary: Genitourinary
Kidney cyst volume (29808.1)	Kidney cyst volume in mL	0..1	volumeInML

Size of left kidney (29809.1)	Left kidney volume in mL	0..1	volumeInML
Size of right kidney (29810.1)	Right kidney volume in mL	0..1	volumeInML
Total kidney volume (29811.1)	Volume of both kidneys in mL	0..1	volumeInML

or, in the case of

3.3.5.4 Liver Imaging (29823.2)

Name	Description	Multiplicity	Data Type
Imaging Modality (34294.1)	Imaging Modality for Liver	1..1	Imaging Procedure (Liver) <i>Enumeration</i> Computerised Axial Tomography: Computerised Axial Tomography Diagnostic Ultrasonography: Diagnostic Ultrasonography Magnetic Resonance Imaging: Magnetic Resonance Imaging Nuclear Medicine: Nuclear Medicine Plain Radiography: Plain Radiography Positron Emission Tomography: Positron Emission Tomography Single Photon Emission Computed Tomography: Single Photon Emission Computed Tomography SAP scan: SAP scan Other: Other
Imaging Submodality (34297.1)	Imaging Submodality for Liver	0..1	Imaging Submodality (Liver) <i>Enumeration</i> DEXA: dual energy X-ray absorptiometry DMSA: dimercaptosuccinic acid scan Ferriscan: Ferriscan

			T2*MRI:T2*MRI
Region (33798.1)	Liver Region	1..1	liverRegion
System (33807.1)	Liver system	1..1	liverSystem
Total liver volume (29824.1)	Total liver volume in mL	0..1	volumeInML
Liver cyst volume (29825.1)	Liver cyst volume in mL	0..1	volumeInML

or, in the case of

3.3.5.5 Facial features most in keeping with an OMIM disease (29826.2)

Name	Description	Multiplicity	Data Type
OMIM Code (29827.1)	OMIM code that best describes disorder	0..1	xs:string
Imaging Modality (33818.1)	Imaging Modality Facial Photograph	1..1	imagingModalityFacial <i>Enumeration</i> Medical Photograph:Medical Photograph
Region (34300.1)	Body region for facial photographs	1..1	Body Region (face) <i>Enumeration</i> Head:Head

or, in the case of

3.3.5.6 Heart/liver Iron measurement (30137.2)

Name	Description	Multiplicity	Data Type
Imaging Modality (34305.1)	Imaging Modality for heart/liver iron measurement	1..1	Imaging Modality (heart/liver) <i>Enumeration</i> Magnetic Resonance Imaging:Magnetic Resonance Imaging Other:Other

Imaging Submodality (34306.1)	Imaging Submodality for heart/liver iron measurement	1..1	Imaging Submodality (heart/liver) <i>Enumeration</i> Ferriscan: Ferriscan T2*MRI: T2*MRI
Body Region (34311.1)	Body region for heart/liver iron measurement	1..1	Body Region (heart/liver) <i>Enumeration</i> Heart: Heart Liver: Liver
Body System (34312.1)	Body system for heart/liver iron measurement	1..1	Body System (heart/liver) <i>Enumeration</i> Cardiovascular: Cardiovascular Digestive: Digestive
Ferriscan result (30142.1)	Measurement of liver iron concentration in g	0..1	mass
T2*MRI liver result interpretation (30149.2)	Interpretation of iron quantification in liver by T2*MRI	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
T2*MRI heart result interpretation (30148.2)	Interpretation of iron quantification in liver by T2*MRI	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.5.6.1 T2*MRI liver (35454.1)

Name	Description	Multiplicity	Data Type
T2*MRI liver result (35451.1)		1..1	timeIntervalMs
T2*MRI liver Upper Range (35452.1)		0..1	timeIntervalMs
T2*MRI liver Lower Range (35453.1)		0..1	timeIntervalMs

together with, in the case of

3.3.5.6.2 T2*MRI heart (35458.1)

Name	Description	Multiplicity	Data Type
T2*MRI heart result (35455.1)		1..1	timeIntervalMs
T2*MRI heart Upper Range (35456.1)		0..1	timeIntervalMs
T2*MRI heart Lower Range (35457.1)		0..1	timeIntervalMs

or, in the case of

3.3.5.7 Cardiac MRI (31429.2)

Name	Description	Multiplicity	Data Type
Imaging Modality (34321.1)	Imaging Modality for cardiac MRI	1..1	Imaging Modality (heart MRI) <i>Enumeration</i> Magnetic Resonance Imaging: Magnetic Resonance Imaging
Imaging Submodality (34322.1)	Imaging Submodality for cardiac MRI	1..1	Imaging Submodality (heart MRI) <i>Enumeration</i> Cardiac MRI: Cardiac MRI
System (34324.1)	Body System for cardiac imaging	1..1	Body System (heart)

			<i>Enumeration</i> Cardiovascular: Cardiovascular
Region (34323.1)	Body region for cardiac imaging	1..1	Body Region (heart) <i>Enumeration</i> Heart: Heart
Structural disease indicated (31430.1)	Structural disease indicated by cardiac MRI	1..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Structural disease description (31431.1)	Description of structural disease indicated by a cardiac MRI investigation	0..1	xs:string

or, in the case of

3.3.5.8 Doppler Diastolic function assessment (29051.2)

Name	Description	Multiplicity	Data Type
Imaging Modality (34327.1)	Imaging Modality for Doppler Diastolic Function Assessment	1..1	Imaging Modality (Doppler Diastolic Function) <i>Enumeration</i> Doppler Diastolic Function Assessment: Doppler Diastolic Function Assessment
System (34324.1)	Body System for cardiac imaging	1..1	Body System (heart) <i>Enumeration</i> Cardiovascular: Cardiovascular
Region (34323.1)	Body region for cardiac imaging	1..1	Body Region (heart) <i>Enumeration</i> Heart: Heart
Doppler Diastolic Function Assessment (31459.1)	Doppler Diastolic Function Assessment	0..1	dopplerDiastolic <i>Enumeration</i> Normal: Normal Type 1: Type 1 Type 2: Type 2

			Type 3: Type 3 Type 4: Type 4
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3.3.6 Laboratory Test Report (29869.2)

All laboratory reports should include the following elements,

Name	Description	Multiplicity	Data Type
Specimen Type Other (30458.1)	If Specimen type is other please specify:	0..1	xs:string
Sampling Preconditions (29876.1)	Indicate any preconditions applicable to sampling, for example fasting status	0..1	samplingPreconditions <i>Enumeration</i> Fasting sample: Post Fasting Sample Post mortem sample: Post Mortem Sample Post-dose sample: Post-dose sample Pre-dose sample: Pre-dose sample Random Sample: Random Sample Sample from ambulatory subject: Sample from ambulatory subject Sample from orthostatic subject: Sample from orthostatic subject Sample from rested subject: Sample from rested subject Sample from subject of unknown posture: Sample from subject of unknown posture Sample from supine subject: Sample from supine subject
Time Aspect (29880.1)	Indicate any time aspect of specimen collection, e.g. spot or 24 hour sampling	0..1	timeAspect <i>Enumeration</i> Spot: Spot Unspecified: Unspecified 24hr Collection: 24hr Collection Basal: Basal - Time Course Stimulated Peak: Stimulated Peak - Time Course

Assessment (29524.1)	Assessment of findings and clinical significance	0..1	<p>Clinical Test Abnormality <i>Enumeration</i></p> <p>Normal:Normal</p> <p>Unknown:No results available</p> <p>Abnormal-Relevant:An abnormality of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>
Patient Status (34004.1)	Represents the patient's status when test was performed	1..1	<p>patientStatus <i>Enumeration</i></p> <p>presenting:Taken at patient presentation</p> <p>diagnostic:Taken at the point of diagnosis</p> <p>baseline:Representing a baseline measurement</p> <p>most abnormal:Representing the most abnormal measurement</p> <p>unknown:Status unknown</p>
Report (29501.2)	File name of uploaded copy of clinical report - requested format [Participant ID]_[Local Report Identifier]	0..1	xs:base64Binary

together with, in the case of

3.3.6.1 General Laboratory Test Report (34329.1)

Name	Description	Multiplicity	Data Type
Analytical technique (29877.2)	If relevant please state physical analytic technique or protocol used for the test.	0..unbounded	<p>labAnalysisType <i>Enumeration</i></p> <p>>10 Enumerations please click above for more details</p>
Specimen Type	Indicated the specimen type used	0..unbounded	analysedSpecimenType <i>Enumeration</i>

(29875.1)	for analysis		>10 Enumerations please click above for more details
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or, in the case of

3.3.6.2 Urine Test (29870.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34483.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrineTest <i>Enumeration</i> Urine Test: Urine Test
Specimen Type (34486.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType UrineTest <i>Enumeration</i> Urine: Urine

together with, in the case of

3.3.6.2.1 Urine electrolytes (33187.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34489.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrineelectrolytes <i>Enumeration</i> Urine electrolytes: Urine electrolytes
Urinary acidification result (30456.2)	ammonium chloride or furosemide and fludrocortisone test	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.2.2 Sodium (34858.1)

Name	Description	Multiplicity	Data Type
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Sodium result (34859.1)	Sodium result	1..1	testResult
Sodium measurement unit (34860.1)	measurement unit used to record Sodium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Sodium Upper Range (34861.1)	upper range of results for Sodium	0..1	testResult
Sodium Lower Range (34862.1)	upper range of results for Sodium	0..1	testResult

together with, in the case of

3.3.6.2.3 Creatinine (34863.1)

Name	Description	Multiplicity	Data Type
Creatinine result (34864.1)	Creatinine result	1..1	testResult
Creatinine measurement unit (34865.1)	measurement unit used to record Creatinine	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Creatinine Upper Range (34866.1)	upper range of results for Creatinine	0..1	testResult
Creatinine Lower Range (34867.1)	upper range of results for Creatinine	0..1	testResult

together with, in the case of

3.3.6.2.4 Potassium (34868.1)

Name	Description	Multiplicity	Data Type
Potassium result (34869.1)	Potassium result	1..1	testResult
Potassium measurement unit (34870.1)	measurement unit used to record Potassium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Potassium Upper Range (34871.1)	upper range of results for Potassium	0..1	testResult
Potassium Lower Range (34872.1)	upper range of results for Potassium	0..1	testResult

together with, in the case of

3.3.6.2.5 Urine Chloride (34883.1)

Name	Description	Multiplicity	Data Type
Urine Chloride result (34884.1)	Urine Chloride result	1..1	testResult
Urine Chloride measurement unit (34885.1)	measurement unit used to record Urine Chloride	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine Chloride Upper Range (34886.1)	upper range of results for Urine Chloride	0..1	testResult
Urine Chloride Lower Range (34887.1)	upper range of results for Urine Chloride	0..1	testResult

together with, in the case of

3.3.6.2.6 Urine Citrate (34888.1)

Name	Description	Multiplicity	Data Type
Urine Citrate result (34889.1)	Urine Citrate result	1..1	testResult
Urine Citrate measurement unit (34890.1)	measurement unit used to record Urine Citrate	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine Citrate Upper Range (34891.1)	upper range of results for Urine Citrate	0..1	testResult
Urine Citrate Lower Range (34892.1)	upper range of results for Urine Citrate	0..1	testResult

together with, in the case of

3.3.6.2.7 Urine Calcium (34893.1)

Name	Description	Multiplicity	Data Type
Urine Calcium result (34894.1)	Urine Calcium result	1..1	testResult
Urine Calcium measurement unit (34895.1)	measurement unit used to record Urine Calcium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click

			<i>above for more details</i>
Urine Calcium Upper Range (34896.1)	upper range of results for Urine Calcium	0..1	testResult
Urine Calcium Lower Range (34897.1)	upper range of results for Urine Calcium	0..1	testResult

together with, in the case of

3.3.6.2.8 Urine calcium / Creatinine excretion ratio (34873.1)

Name	Description	Multiplicity	Data Type
Urine calcium / Creatinine excretion ratio measurement result (34874.1)	Urine calcium / Creatinine excretion ratio result	1..1	testResult
Urine calcium / Creatinine excretion ratio measurement Upper Range (34876.1)	upper range of results for Urine calcium / Creatinine excretion ratio measurement	0..1	testResult
Urine calcium / Creatinine excretion ratio measurement Lower Range (34877.1)	upper range of results for Urine calcium / Creatinine excretion ratio measurement	0..1	testResult

together with, in the case of

3.3.6.2.9 Urine osmolality (35418.1)

Name	Description	Multiplicity	Data Type
Urine osmolality result (35414.1)		1..1	xs:decimal
Urine osmolality measurement unit (35415.1)		1..1	concentrationUnitOfMeasurement <i>Enumeration</i> <i>>10 Enumerations please click above for more details</i>
Urine osmolality Upper Range (35416.1)		0..1	xs:decimal
Urine osmolality Lower Range (35417.1)		0..1	xs:decimal

together with, in the case of

3.3.6.2.10 Urine Magnesium (34898.1)

Name	Description	Multiplicity	Data Type
Urine Magnesium result (34899.1)	Urine Magnesium result	1..1	testResult
Urine Magnesium measurement unit (34900.1)	measurement unit used to record Urine Magnesium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i>
Urine Magnesium Upper Range (34901.1)	upper range of results for Urine Magnesium	0..1	testResult
Urine Magnesium Lower Range (34902.1)	upper range of results for Urine Magnesium	0..1	testResult

together with, in the case of

3.3.6.2.11 Urine pH (34903.1)

Name	Description	Multiplicity	Data Type
Urine pH result (34904.1)	Urine pH result	1..1	testResult
Urine pH Upper Range (34906.1)	upper range of results for Urine pH	0..1	testResult
Urine pH Lower Range (34907.1)	upper range of results for Urine pH	0..1	testResult

together with, in the case of

3.3.6.2.12 Urine Phosphate (34908.1)

Name	Description	Multiplicity	Data Type
Urine Phosphate result (34909.1)	Urine Phosphate result	1..1	testResult
Urine Phosphate measurement unit (34910.1)	measurement unit used to record Urine Phosphate	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine Phosphate Upper Range (34911.1)	upper range of results for Urine Phosphate	0..1	testResult

Urine Phosphate Lower Range (34912.1)	upper range of results for Urine Phosphate	0..1	testResult
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together with, in the case of

3.3.6.2.13 Urine glucose (33188.1).

Name	Description	Multiplicity	Data Type
Analytical technique (34495.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrineglucose <i>Enumeration</i> Urine glucose: Urine glucose

together with, in the case of

3.3.6.2.14 Urine Glucose (34913.1)

Name	Description	Multiplicity	Data Type
Urine Glucose result (34914.1)	Urine Glucose result	1..1	testResult
Urine Glucose measurement unit (34915.1)	measurement unit used to record Urine Glucose	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine Glucose Upper Range (34916.1)	upper range of results for Urine Glucose	0..1	testResult
Urine Glucose Lower Range (34917.1)	upper range of results for Urine Glucose	0..1	testResult

together with, in the case of

3.3.6.2.15 Urine metabolic tests (33189.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34501.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrinemetabolictests <i>Enumeration</i> Urine metabolic tests: Urine metabolic tests
Urinary amino acids result (28785.2)	Results of Urinary amino acid test	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality

			<p>of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>
Urine organic acids (33286.1)	Urine organic acid testing result	0..1	<p>Clinical Test Abnormality <i>Enumeration</i></p> <p>Normal:Normal</p> <p>Unknown:No results available</p> <p>Abnormal-Relevant:An abnormality of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>
Glycosaminoglycans result (30445.2)	Glycosaminoglycans	0..1	<p>Clinical Test Abnormality <i>Enumeration</i></p> <p>Normal:Normal</p> <p>Unknown:No results available</p> <p>Abnormal-Relevant:An abnormality of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>
Urine oligosaccharides result (30448.2)	Urine oligosaccharides	0..1	<p>Clinical Test Abnormality <i>Enumeration</i></p> <p>Normal:Normal</p> <p>Unknown:No results available</p> <p>Abnormal-Relevant:An abnormality of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>
Urine Purines result (30453.2)	xanthine and hypoxanthine	0..1	<p>Clinical Test Abnormality <i>Enumeration</i></p> <p>Normal:Normal</p> <p>Unknown:No results available</p> <p>Abnormal-Relevant:An abnormality of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown Relevance:An abnormality of unknown clinical</p>

			relevance to the patient's condition
Urine succinylacetone result (30455.2)	Urine succinylacetone	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.2.16 Urine copper (34918.1)

Name	Description	Multiplicity	Data Type
Urine copper result (34919.1)	Urine copper result	1..1	testResult
Urine copper measurement unit (34920.1)	measurement unit used to record Urine copper	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine copper Upper Range (34921.1)	upper range of results for Urine copper	0..1	testResult
Urine copper Lower Range (34922.1)	upper range of results for Urine copper	0..1	testResult

together with, in the case of

3.3.6.2.17 Urine caeruloplasmin (34923.1)

Name	Description	Multiplicity	Data Type
Urine caeruloplasmin result (34924.1)	Urine caeruloplasmin result	1..1	testResult
Urine caeruloplasmin measurement unit (34925.1)	measurement unit used to record Urine caeruloplasmin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine caeruloplasmin Upper Range (34926.1)	upper range of results for Urine caeruloplasmin	0..1	testResult

Urine caeruloplasmin Lower Range (34927.1)	upper range of results for Urine caeruloplasmin	0..1	testResult
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together with, in the case of

3.3.6.2.18 Urine protein (33190.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34507.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrineprotein <i>Enumeration</i> Urine protein: Urine protein
Low molecular weight protein result (30446.1)	retinol binding protein or β 2-microglobulin	0..1	lowMolecularWeightProtein <i>Enumeration</i> High: Raised above normal level Low: Reduced below normal level Normal: At a normal level

together with, in the case of

3.3.6.2.19 Albumin creatinine ratio (34928.1)

Name	Description	Multiplicity	Data Type
Albumin creatinine ratio result (34929.1)	Albumin creatinine ratio result	1..1	testResult
Albumin creatinine ratio Upper Range (34931.1)	upper range of results for Albumin creatinine ratio	0..1	testResult
Albumin creatinine ratio Lower Range (34932.1)	upper range of results for Albumin creatinine ratio	0..1	testResult

together with, in the case of

3.3.6.2.20 Urine albumin creatinine ratio (34933.1)

Name	Description	Multiplicity	Data Type
Urine albumin creatinine ratio result (34934.1)	Urine albumin creatinine ratio result	1..1	testResult
Urine albumin	upper range of results for Urine	0..1	testResult

creatinine ratio Upper Range (34936.1)	albumin creatinine ratio		
Urine albumin creatinine ratio Lower Range (34937.1)	upper range of results for Urine albumin creatinine ratio	0..1	testResult

together with, in the case of

3.3.6.2.21 Urine albumin Level (34938.1)

Name	Description	Multiplicity	Data Type
Urine albumin Level result (34939.1)	Urine albumin Level result	1..1	testResult
Urine albumin Level measurement unit (34940.1)	measurement unit used to record Urine albumin Level	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine albumin Level Upper Range (34941.1)	upper range of results for Urine albumin Level	0..1	testResult
Urine albumin Level Lower Range (34942.1)	upper range of results for Urine albumin Level	0..1	testResult

together with, in the case of

3.3.6.2.22 Urine Protein Creatinine ratio (34943.1)

Name	Description	Multiplicity	Data Type
Urine Protein Creatinine ratio result (34944.1)	Urine Protein Creatinine ratio result	1..1	testResult
Urine Protein Creatinine ratio Upper Range (34946.1)	upper range of results for Urine Protein Creatinine ratio	0..1	testResult
Urine Protein Creatinine ratio Lower Range (34947.1)	upper range of results for Urine Protein Creatinine ratio	0..1	testResult

together with, in the case of

3.3.6.2.23 Urine Protein (34948.1)

Name	Description	Multiplicity	Data Type
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Urine Protein result (34949.1)	Urine Protein result	1..1	testResult
Urine Protein measurement unit (34950.1)	measurement unit used to record Urine Protein	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Urine Protein Upper Range (34951.1)	upper range of results for Urine Protein	0..1	testResult
Urine Protein Lower Range (34952.1)	upper range of results for Urine Protein	0..1	testResult

together with, in the case of

3.3.6.2.24 Urine reducing substances (33191.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34513.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrinereducingsubstances <i>Enumeration</i> Urine reducing substances: Urine reducing substances
Urinary reducing substances (e.g. galactosaemia) present (28782.1)	Urinary reducing substances (e.g. galactosaemia) present?	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown

or, in the case of

3.3.6.3 Urine Dip (29871.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34519.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrineDip <i>Enumeration</i> Urine Dip: Urine Dip
Specimen Type (34522.1)	Indicated the specimen type used for analysis	0..unbounded	analysedSpecimenType UrineDip <i>Enumeration</i> Urine: Urine

together with, in the case of together with, in the case of

3.3.6.3.1 Urine dip - standard (31317.2).

Name	Description	Multiplicity	Data Type
Analytical technique (34525.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrinedip-standard <i>Enumeration</i> Urine dip - standard: Urine dip - standard
Leucocyte dip result (16371.2)	URINE DIP result	0..1	leucocyteDipResult <i>Enumeration</i> Negative: Negative Trace: Trace Small: Small Moderate: Moderate Large: Large
Nitrite dip result (16373.2)	URINE DIP nitrite result	0..1	PositiveNegativeUnknown <i>Enumeration</i> unknown: Unknown negative: Negative positive: Positive
Glucose dip result (16379.1)	URINE DIP glucose result	0..1	glucoseDipResult
Protein dip result (16377.2)	URINE DIP protein result	0..1	concentrationMmol/l
Ketone bodies dip result (16381.2)	URINE DIP ketone bodies result	0..1	concentrationMmol/l
Hb dip result (16385.2)	URINE DIP Hb result	0..1	hbDipResult <i>Enumeration</i> Negative: Negative Non-haemolysed trace: Non-haemolysed trace Haemolysed trace: Haemolysed trace Small: Small Moderate: Moderate Large: Large
pH dip result (16375.1)	URINE DIP pH result	0..1	xs:string

together with, in the case of

3.3.6.3.2 Urine dip - sulphites (31318.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34531.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeUrineDipSulphites <i>Enumeration</i> Urine dip - sulphites: Urine dip - sulphites
Sulphites Dip Result (30303.2)	URINE DIP sulphite result	0..1	PositiveNegativeUnknown <i>Enumeration</i> unknown: Unknown negative: Negative positive: Positive

or, in the case of

3.3.6.4 Culture (28285.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34537.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeCulture <i>Enumeration</i> Culture: Culture
Specimen Type (29875.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType <i>Enumeration</i> >10 Enumerations please click above for more details
Site (28286.1)	Microbiology Culture Site measurement. Recorded 1:1	0..1	xs:string
Organism (28287.1)	Microbiology Culture organism measurement. Recorded 1:1	0..1	xs:string
Sensitivity (28288.1)	Microbiology Culture sensitivity measurement. Recorded 1:1	0..1	xs:string
Date and time of microbiology culture measurement (28289.1)	Date and time of microbiology culture measurement	0..1	xs:dateTime

or, in the case of

3.3.6.5 Autoantibodies (30112.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34543.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeAutoantibodies <i>Enumeration</i> Autoantibodies: Autoantibodies
Specimen Type (34546.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Autoantibodies <i>Enumeration</i> Blood: Blood

together with a set of autoantibody results,

3.3.6.5.1 Autoantibody (31150.2)

Name	Description	Multiplicity	Data Type
Antibody Result (30121.1)	The result of the antibody test	1..1	antibodyResult <i>Enumeration</i> positive: Positive negative: Negative unknown: Unknown significance not done: Not done
Antibody type (30113.2)	The specific antibody type	1..1	antibodyName <i>Enumeration</i> >10 Enumerations please click above for more details

or, in the case of

3.3.6.6 TORCH screen (30122.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34555.1)	If relevant please state physical analytic technique or protocol	1..1	labAnalysisTypeTORCHscreen <i>Enumeration</i>

	used for the test.		TORCH screen: TORCH screen
Specimen Type (34558.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType TORCHscreen <i>Enumeration</i> Blood: Blood
Toxoplasmosis IgG (33376.1)		0..1	antibodyResult <i>Enumeration</i> positive: Positive negative: Negative unknown: Unknown significance not done: Not done
Toxoplasmosis IgM (33375.1)		0..1	antibodyResult <i>Enumeration</i> positive: Positive negative: Negative unknown: Unknown significance not done: Not done
Rubella IgM (33377.1)		0..1	antibodyResult <i>Enumeration</i> positive: Positive negative: Negative unknown: Unknown significance not done: Not done
Herpes simplex virus IgM (33381.1)		0..1	antibodyResult <i>Enumeration</i> positive: Positive negative: Negative unknown: Unknown significance not done: Not done
Herpes simplex virus IgG (33382.1)		0..1	antibodyResult <i>Enumeration</i> positive: Positive negative: Negative unknown: Unknown significance not done: Not done

Cytomegalovirus IgM (33379.1)		0..1	antibodyResult <i>Enumeration</i> positive :Positive negative :Negative unknown :Unknown significance not done :Not done
Cytomegalovirus IgG (33380.1)		0..1	antibodyResult <i>Enumeration</i> positive :Positive negative :Negative unknown :Unknown significance not done :Not done
Rubella IgG (33378.1)		0..1	antibodyResult <i>Enumeration</i> positive :Positive negative :Negative unknown :Unknown significance not done :Not done
HIV IgM (33383.1)		0..1	antibodyResult <i>Enumeration</i> positive :Positive negative :Negative unknown :Unknown significance not done :Not done
HIV IgG (33384.1)		0..1	antibodyResult <i>Enumeration</i> positive :Positive negative :Negative unknown :Unknown significance not done :Not done

or, in the case of

3.3.6.7 Arterial blood gas (30304.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34561.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeArterialbloodgas <i>Enumeration</i> Arterial blood gas: Arterial blood gas
Specimen Type (34564.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Arterialbloodgas <i>Enumeration</i> Blood: Blood

together with, in the case of

3.3.6.7.1 HCO₃ - ABG / VBG (34953.1)

Name	Description	Multiplicity	Data Type
HCO₃ - ABG / VBG result (34954.1)	HCO ₃ - ABG / VBG result	1..1	testResult
HCO₃ - ABG / VBG measurement unit (34955.1)	measurement unit used to record HCO ₃ - ABG / VBG	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
HCO₃ - ABG / VBG Upper Range (34956.1)	upper range of results for HCO ₃ - ABG / VBG	0..1	testResult
HCO₃ - ABG / VBG Lower Range (34957.1)	upper range of results for HCO ₃ - ABG / VBG	0..1	testResult

together with, in the case of

3.3.6.7.2 Lactate - ABG (34958.1)

Name	Description	Multiplicity	Data Type
Lactate - ABG result (34959.1)	Lactate - ABG result	1..1	testResult
Lactate - ABG measurement unit (34960.1)	measurement unit used to record Lactate - ABG	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Lactate - ABG Upper Range (34961.1)	upper range of results for Lactate - ABG	0..1	testResult

Lactate - ABG Lower Range (34962.1)	upper range of results for Lactate - ABG	0..1	testResult
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together with, in the case of

3.3.6.7.3 pH - ABG / VBG (34963.1)

Name	Description	Multiplicity	Data Type
pH - ABG / VBG result (34964.1)	pH - ABG / VBG result	1..1	testResult
pH - ABG / VBG measurement unit (34965.1)	measurement unit used to record pH - ABG / VBG	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
pH - ABG / VBG Upper Range (34966.1)	upper range of results for pH - ABG / VBG	0..1	testResult
pH - ABG / VBG Lower Range (34967.1)	upper range of results for pH - ABG / VBG	0..1	testResult

together with, in the case of

3.3.6.7.4 Sodium ABG/VBG (34968.1)

Name	Description	Multiplicity	Data Type
Sodium ABG/VBG result (34969.1)	Sodium ABG/VBG result	1..1	testResult
Sodium ABG/VBG measurement unit (34970.1)	measurement unit used to record Sodium ABG/VBG	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Sodium ABG/VBG Upper Range (34971.1)	upper range of results for Sodium ABG/VBG	0..1	testResult
Sodium ABG/VBG Lower Range (34972.1)	upper range of results for Sodium ABG/VBG	0..1	testResult

together with, in the case of

3.3.6.7.5 Potassium ABG/VBG (34973.1)

Name	Description	Multiplicity	Data Type
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Potassium ABG/VBG result (34974.1)	Potassium ABG/VBG result	1..1	testResult
Potassium ABG/VBG measurement unit (34975.1)	measurement unit used to record Potassium ABG/VBG	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Potassium ABG/VBG Upper Range (34976.1)	upper range of results for Potassium ABG/VBG	0..1	testResult
Potassium ABG/VBG Lower Range (34977.1)	upper range of results for Potassium ABG/VBG	0..1	testResult

or, in the case of

3.3.6.8 Bone profile (30317.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34567.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeBoneprofile <i>Enumeration</i> Bone profile: Bone profile
Specimen Type (34570.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Boneprofile <i>Enumeration</i> Blood: Blood

together with, in the case of

3.3.6.8.1 Calcium (34978.1)

Name	Description	Multiplicity	Data Type
Calcium result (34979.1)	Calcium result	1..1	testResult
Calcium measurement unit (34980.1)	measurement unit used to record Calcium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Calcium Upper Range (34981.1)	upper range of results for Calcium	0..1	testResult

Calcium Lower Range (34982.1)	upper range of results for Calcium	0..1	testResult
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together with, in the case of

3.3.6.8.2 Corrected Calcium (34983.1)

Name	Description	Multiplicity	Data Type
Corrected Calcium result (34984.1)	Corrected Calcium result	1..1	testResult
Corrected Calcium measurement unit (34985.1)	measurement unit used to record Corrected Calcium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Corrected Calcium Upper Range (34986.1)	upper range of results for Corrected Calcium	0..1	testResult
Corrected Calcium Lower Range (34987.1)	upper range of results for Corrected Calcium	0..1	testResult

together with, in the case of

3.3.6.8.3 Total protein level (34988.1)

Name	Description	Multiplicity	Data Type
Total protein level result (34989.1)	Total protein level result	1..1	testResult
Total protein level measurement unit (34990.1)	measurement unit used to record Total protein level	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Total protein level Upper Range (34991.1)	upper range of results for Total protein level	0..1	testResult
Total protein level Lower Range (34992.1)	upper range of results for Total protein level	0..1	testResult

together with, in the case of

3.3.6.8.4 Albumin (34993.1)

Name	Description	Multiplicity	Data Type
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Albumin result (34994.1)	Albumin result	1..1	testResult
Albumin measurement unit (34995.1)	measurement unit used to record Albumin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Albumin Upper Range (34996.1)	upper range of results for Albumin	0..1	testResult
Albumin Lower Range (34997.1)	upper range of results for Albumin	0..1	testResult

together with, in the case of

3.3.6.8.5 Alkaline Phosphatase (34998.1)

Name	Description	Multiplicity	Data Type
Alkaline Phosphatase result (34999.1)	Alkaline Phosphatase result	1..1	testResult
Alkaline Phosphatase measurement unit (35000.1)	measurement unit used to record Alkaline Phosphatase	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Alkaline Phosphatase Upper Range (35001.1)	upper range of results for Alkaline Phosphatase	0..1	testResult
Alkaline Phosphatase Lower Range (35002.1)	upper range of results for Alkaline Phosphatase	0..1	testResult

together with, in the case of

3.3.6.8.6 Phosphate (35003.1)

Name	Description	Multiplicity	Data Type
Phosphate result (35004.1)	Phosphate result	1..1	testResult
Phosphate measurement unit (35005.1)	measurement unit used to record Phosphate	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details

Phosphate Upper Range (35006.1)	upper range of results for Phosphate	0..1	testResult
Phosphate Lower Range (35007.1)	upper range of results for Phosphate	0..1	testResult

together with, in the case of

3.3.6.8.7 Parathyroid hormone measurement (35008.1)

Name	Description	Multiplicity	Data Type
Parathyroid hormone measurement result (35009.1)	Parathyroid hormone measurement result	1..1	testResult
Parathyroid hormone measurement measurement unit (35010.1)	measurement unit used to record Parathyroid hormone measurement	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Parathyroid hormone measurement Upper Range (35011.1)	upper range of results for Parathyroid hormone measurement	0..1	testResult
Parathyroid hormone measurement Lower Range (35012.1)	upper range of results for Parathyroid hormone measurement	0..1	testResult

3.3.6.9 Full Blood Count (30318.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34573.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeFullBloodCount <i>Enumeration</i> Full Blood Count: Full Blood Count
Specimen Type (34576.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType FullBloodCount <i>Enumeration</i> Blood: Blood
Blood film result (30383.2)	Blood film	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available

			Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
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together with, in the case of

3.3.6.9.1 Haemoglobin (35013.1)

Name	Description	Multiplicity	Data Type
HAEMOGLOBIN result (35014.1)	HAEMOGLOBIN result	1..1	testResult
HAEMOGLOBIN measurement unit (35015.1)	measurement unit used to record HAEMOGLOBIN	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x109/l)
HAEMOGLOBIN Upper Range (35016.1)	upper range of results for HAEMOGLOBIN	0..1	testResult
HAEMOGLOBIN Lower Range (35017.1)	upper range of results for HAEMOGLOBIN	0..1	testResult

together with, in the case of

3.3.6.9.2 Haematocrit determination (35018.1)

Name	Description	Multiplicity	Data Type
Haematocrit determination result (35019.1)	Haematocrit determination result	1..1	testResult
Haematocrit determination measurement unit (35020.1)	measurement unit used to record Haematocrit determination	1..1	numberUnitOfMeasurement <i>Enumeration</i> Number (Retired September 2013): Number (Retired September 2013) Percentage (%): Percentage

			(%)
Haematocrit determination Upper Range (35021.1)	upper range of results for Haematocrit determination	0..1	testResult
Haematocrit determination Lower Range (35022.1)	upper range of results for Haematocrit determination	0..1	testResult

together with, in the case of

3.3.6.9.3 Red Blood Cell count (35023.1)

Name	Description	Multiplicity	Data Type
Red Blood Cell count result (35024.1)	Red Blood Cell count result	1..1	testResult
Red Blood Cell count measurement unit (35025.1)	measurement unit used to record Red Blood Cell count	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x10 ⁹ /l)
Red Blood Cell count Upper Range (35026.1)	upper range of results for Red Blood Cell count	0..1	testResult
Red Blood Cell count Lower Range (35027.1)	upper range of results for Red Blood Cell count	0..1	testResult

together with, in the case of

3.3.6.9.4 Red cell width distribution determination (35028.1)

Name	Description	Multiplicity	Data Type
Red cell width distribution determination result (35029.1)	Red cell width distribution determination result	1..1	testResult
Red cell width distribution determination	measurement unit used to record Red cell width distribution determination	1..1	numberUnitOfMeasurement <i>Enumeration</i> Number (Retired)

measurement unit (35030.1)			September 2013): Number (Retired September 2013) Percentage (%): Percentage (%)
Red cell width distribution determination Upper Range (35031.1)	upper range of results for Red cell width distribution determination	0..1	testResult
Red cell width distribution determination Lower Range (35032.1)	upper range of results for Red cell width distribution determination	0..1	testResult

together with, in the case of

3.3.6.9.5 Mean cell haemoglobin (MCH) (35033.1)

Name	Description	Multiplicity	Data Type
Mean cell haemoglobin (MCH) result (35034.1)	Mean cell haemoglobin (MCH) result	1..1	testResult
Mean cell haemoglobin (MCH) measurement unit (35035.1)	measurement unit used to record Mean cell haemoglobin (MCH)	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x10 ⁹ /l)
Mean cell haemoglobin (MCH) Upper Range (35036.1)	upper range of results for Mean cell haemoglobin (MCH)	0..1	testResult
Mean cell haemoglobin (MCH) Lower Range (35037.1)	upper range of results for Mean cell haemoglobin (MCH)	0..1	testResult

together with, in the case of

3.3.6.9.6 Mean Cell Volume (MCV) (35038.1)

Name	Description	Multiplicity	Data Type
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Mean Cell Volume (MCV) result (35039.1)	Mean Cell Volume (MCV) result	1..1	testResult
Mean Cell Volume (MCV) measurement unit (35040.1)	measurement unit used to record Mean Cell Volume (MCV)	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x10 ⁹ /l)
Mean Cell Volume (MCV) Upper Range (35041.1)	upper range of results for Mean Cell Volume (MCV)	0..1	testResult
Mean Cell Volume (MCV) Lower Range (35042.1)	upper range of results for Mean Cell Volume (MCV)	0..1	testResult

together with, in the case of

3.3.6.9.7 White Blood Cell count (35043.1)

Name	Description	Multiplicity	Data Type
White Blood Cell count result (35044.1)	White Blood Cell count result	1..1	testResult
White Blood Cell count measurement unit (35045.1)	measurement unit used to record White Blood Cell count	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x10 ⁹ /l)
White Blood Cell count Upper Range (35046.1)	upper range of results for White Blood Cell count	0..1	testResult
White Blood Cell count Lower Range (35047.1)	upper range of results for White Blood Cell count	0..1	testResult

together with, in the case of

3.3.6.9.8 Neutrophils (35048.1)

Name	Description	Multiplicity	Data Type
Neutrophils result (35049.1)	Neutrophils result	1..1	testResult
Neutrophils measurement unit (35050.1)	measurement unit used to record Neutrophils	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l) :number times ten raised to the power of nine per litre (x10 ⁹ /l)
Neutrophils Upper Range (35051.1)	upper range of results for Neutrophils	0..1	testResult
Neutrophils Lower Range (35052.1)	upper range of results for Neutrophils	0..1	testResult

together with, in the case of

3.3.6.9.9 Lymphocytes (35053.1)

Name	Description	Multiplicity	Data Type
Lymphocytes result (35054.1)	Lymphocytes result	1..1	testResult
Lymphocytes measurement unit (35055.1)	measurement unit used to record Lymphocytes	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l) :number times ten raised to the power of nine per litre (x10 ⁹ /l)
Lymphocytes Upper Range (35056.1)	upper range of results for Lymphocytes	0..1	testResult
Lymphocytes Lower Range (35057.1)	upper range of results for Lymphocytes	0..1	testResult

together with, in the case of

3.3.6.9.10 Monocytes (35058.1)

Name	Description	Multiplicity	Data Type
Monocytes result (35059.1)	Monocytes result	1..1	testResult
Monocytes measurement unit (35060.1)	measurement unit used to record Monocytes	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l) :number times ten raised to the power of nine per litre (x109/l)
Monocytes Upper Range (35061.1)	upper range of results for Monocytes	0..1	testResult
Monocytes Lower Range (35062.1)	upper range of results for Monocytes	0..1	testResult

together with, in the case of

3.3.6.9.11 Eosinophils (35063.1)

Name	Description	Multiplicity	Data Type
Eosinophils result (35064.1)	Eosinophils result	1..1	testResult
Eosinophils measurement unit (35065.1)	measurement unit used to record Eosinophils	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l) :number times ten raised to the power of nine per litre (x109/l)
Eosinophils Upper Range (35066.1)	upper range of results for Eosinophils	0..1	testResult
Eosinophils Lower Range (35067.1)	upper range of results for Eosinophils	0..1	testResult

together with, in the case of

3.3.6.9.12 Basophils (35433.1)

Name	Description	Multiplicity	Data Type
Basophils result (35429.1)		1..1	xs:decimal

Basophils measurement unit (35430.1)		1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x109/l)
Basophils Lower Range (35432.1)		0..1	xs:decimal
Basophils Upper Range (35431.1)		0..1	xs:decimal

together with, in the case of

3.3.6.9.13 Platelets (35438.1)

Name	Description	Multiplicity	Data Type
Platelets result (35434.1)		1..1	xs:decimal
Platelets measurement unit (35435.1)		1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l): number times ten raised to the power of nine per litre (x109/l)
Platelets Upper Range (35436.1)		0..1	xs:decimal
Platelets Lower Range (35437.1)		0..1	xs:decimal

together with, in the case of

3.3.6.9.14 Mean Platelet Volume determination (35073.1)

Name	Description	Multiplicity	Data Type
Mean Platelet Volume determination result (35074.1)	Mean Platelet Volume determination result	1..1	testResult
Mean Platelet Volume determination	measurement unit used to record Mean Platelet Volume determination	1..1	volumeUnitOfMeasurement <i>Enumeration</i> Cubic Millimetres

measurement unit (35075.1)			(mm3) :Cubic Millimetres (mm3) Decilitres (dl) :Decilitres (dl) Femtolitres (fl) :Femtolitres (fl) Litres (l) :Litres (l) Millilitres (ml) :Millilitres (ml) Millilitres (ml) (Retired September 2013) :Millilitres (ml) (Retired September 2013) Nanograms per litre (ng/l) :Nanograms per litre (ng/l)
Mean Platelet Volume determination Upper Range (35076.1)	upper range of results for Mean Platelet Volume determination	0..1	testResult
Mean Platelet Volume determination Lower Range (35077.1)	upper range of results for Mean Platelet Volume determination	0..1	testResult

together with, in the case of

3.3.6.9.15 Reticulocyte count (35068.1)

Name	Description	Multiplicity	Data Type
Reticulocyte count result (35069.1)	Reticulocyte count result	1..1	testResult
Reticulocyte count measurement unit (35070.1)	measurement unit used to record Reticulocyte count	1..1	cellCountUnitOfMeasurement <i>Enumeration</i> number times ten raised to the power of nine per litre (x10⁹/l) :number times ten raised to the power of nine per litre (x10 ⁹ /l)
Reticulocyte count Upper Range (35071.1)	upper range of results for Reticulocyte count	0..1	testResult
Reticulocyte count Lower Range	upper range of results for	0..1	testResult

(35072.1)	Reticulocyte count		
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or, in the case of

3.3.6.10 Liver biochemistry (30328.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34579.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeLiverbiochemistry <i>Enumeration</i> Liver biochemistry: Liver biochemistry
Specimen Type (34582.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Liverbiochemistry <i>Enumeration</i> Blood: Blood

together with, in the case of

3.3.6.10.1 Alanine aminotransferase (ALT) (35078.1)

Name	Description	Multiplicity	Data Type
Alanine aminotransferase (ALT) result (35079.1)	Alanine aminotransferase (ALT) result	1..1	testResult
Alanine aminotransferase (ALT) measurement unit (35080.1)	measurement unit used to record Alanine aminotransferase (ALT)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Alanine aminotransferase (ALT) Upper Range (35081.1)	upper range of results for Alanine aminotransferase (ALT)	0..1	testResult
Alanine aminotransferase (ALT) Lower Range (35082.1)	upper range of results for Alanine aminotransferase (ALT)	0..1	testResult

together with, in the case of

3.3.6.10.2 Albumin (35083.1)

Name	Description	Multiplicity	Data Type
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Albumin result (35084.1)	Albumin result	1..1	testResult
Albumin measurement unit (35085.1)	measurement unit used to record Albumin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Albumin Upper Range (35086.1)	upper range of results for Albumin	0..1	testResult
Albumin Lower Range (35087.1)	upper range of results for Albumin	0..1	testResult

together with, in the case of

3.3.6.10.3 Alkaline phosphatase (35088.1)

Name	Description	Multiplicity	Data Type
Alkaline phosphatase result (35089.1)	Alkaline phosphatase result	1..1	testResult
Alkaline phosphatase measurement unit (35090.1)	measurement unit used to record Alkaline phosphatase	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Alkaline phosphatase Upper Range (35091.1)	upper range of results for Alkaline phosphatase	0..1	testResult
Alkaline phosphatase Lower Range (35092.1)	upper range of results for Alkaline phosphatase	0..1	testResult

together with, in the case of

3.3.6.10.4 Aspartate aminotransferase (AST) (35093.1)

Name	Description	Multiplicity	Data Type
Aspartate aminotransferase (AST) result (35094.1)	Aspartate aminotransferase (AST) result	1..1	testResult
Aspartate aminotransferase (AST) measurement unit (35095.1)	measurement unit used to record Aspartate aminotransferase (AST)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Aspartate	upper range of results for	0..1	testResult

aminotrasferase (AST) Upper Range (35096.1)	Aspartate aminotrasferase (AST)		
Aspartate aminotrasferase (AST) Lower Range (35097.1)	upper range of results for Aspartate aminotrasferase (AST)	0..1	testResult

together with, in the case of

3.3.6.10.5 Bilirubin (total) (35098.1)

Name	Description	Multiplicity	Data Type
Bilirubin (total) result (35099.1)	Bilirubin (total) result	1..1	testResult
Bilirubin (total) measurement unit (35100.1)	measurement unit used to record Bilirubin (total)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Bilirubin (total) Upper Range (35101.1)	upper range of results for Bilirubin (total)	0..1	testResult
Bilirubin (total) Lower Range (35102.1)	upper range of results for Bilirubin (total)	0..1	testResult

together with, in the case of

3.3.6.10.6 Gamma-glutamyl transferase (GGT) (35103.1)

Name	Description	Multiplicity	Data Type
Gamma-glutamyl transferase (GGT) result (35104.1)	Gamma-glutamyl transferase (GGT) result	1..1	testResult
Gamma-glutamyl transferase (GGT) measurement unit (35105.1)	measurement unit used to record Gamma-glutamyl transferase (GGT)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Gamma-glutamyl transferase (GGT) Upper Range (35106.1)	upper range of results for Gamma-glutamyl transferase (GGT)	0..1	testResult
Gamma-glutamyl transferase (GGT) Lower Range	upper range of results for Gamma-glutamyl transferase	0..1	testResult

(35107.1)	(GGT)		
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together with, in the case of

3.3.6.10.7 Total Protein (35108.1)

Name	Description	Multiplicity	Data Type
Total Protein result (35109.1)	Total Protein result	1..1	testResult
Total Protein measurement unit (35110.1)	measurement unit used to record Total Protein	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Total Protein Upper Range (35111.1)	upper range of results for Total Protein	0..1	testResult
Total Protein Lower Range (35112.1)	upper range of results for Total Protein	0..1	testResult

together with, in the case of

3.3.6.10.8 Bilirubin (Direct) (35113.1)

Name	Description	Multiplicity	Data Type
Bilirubin (Direct) result (35114.1)	Bilirubin (Direct) result	1..1	testResult
Bilirubin (Direct) measurement unit (35115.1)	measurement unit used to record Bilirubin (Direct)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Bilirubin (Direct) Upper Range (35116.1)	upper range of results for Bilirubin (Direct)	0..1	testResult
Bilirubin (Direct) Lower Range (35117.1)	upper range of results for Bilirubin (Direct)	0..1	testResult

together with, in the case of

3.3.6.10.9 Bilirubin (Indirect) (35118.1)

Name	Description	Multiplicity	Data Type
Bilirubin (Indirect) result (35119.1)	Bilirubin (Indirect) result	1..1	testResult

Bilirubin (Indirect) measurement unit (35120.1)	measurement unit used to record Bilirubin (Indirect)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Bilirubin (Indirect) Upper Range (35121.1)	upper range of results for Bilirubin (Indirect)	0..1	testResult
Bilirubin (Indirect) Lower Range (35122.1)	upper range of results for Bilirubin (Indirect)	0..1	testResult

or, in the case of

3.3.6.11 Pancreatic autoantibodies (30337.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34585.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypePancreaticautoantibodies <i>Enumeration</i> Pancreatic autoantibodies: Pancreatic autoantibodies
Specimen Type (34588.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Pancreaticautoantibodies <i>Enumeration</i> Blood: Blood
Pancreatic autoantibodies - GAD (28697.1)	Are GAD autoantibodies present?	0..1	PresentAbsent <i>Enumeration</i> Present: Present Absent: Absent
Pancreatic autoantibodies - IA2 (28704.1)	Are IA2 antibodies present?	0..1	PresentAbsent <i>Enumeration</i> Present: Present Absent: Absent
Pancreatic autoantibodies - ICA (28705.1)	Are ICA autoantibodies present?	0..1	PresentAbsent <i>Enumeration</i> Present: Present Absent: Absent
Pancreatic autoantibodies - ZnT8 (28706.1)	Are ZnT8 autoantibodies present?	0..1	PresentAbsent <i>Enumeration</i> Present: Present Absent: Absent

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or, in the case of

3.3.6.12 Serum immunoglobulins (30338.2)

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Name	Description	Multiplicity	Data Type
Analytical technique (34591.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeSerumimmunoglobulins <i>Enumeration</i> Serum immunoglobulins: Serum immunoglobulins
Specimen Type (34594.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Serumimmunoglobulins <i>Enumeration</i> Blood: Blood

together with, in the case of

3.3.6.12.1 Immunoglobulin A (IgA) (35123.1)

Name	Description	Multiplicity	Data Type
Immunoglobulin A (IgA) result (35124.1)	Immunoglobulin A (IgA) result	1..1	testResult
Immunoglobulin A (IgA) measurement unit (35125.1)	measurement unit used to record Immunoglobulin A (IgA)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Immunoglobulin A (IgA) Upper Range (35126.1)	upper range of results for Immunoglobulin A (IgA)	0..1	testResult
Immunoglobulin A (IgA) Lower Range (35127.1)	upper range of results for Immunoglobulin A (IgA)	0..1	testResult

together with, in the case of

3.3.6.12.2 Immunoglobulin G (IgG) (35128.1)

Name	Description	Multiplicity	Data Type
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Immunoglobulin G (IgG) result (35129.1)	Immunoglobulin G (IgG) result	1..1	testResult
Immunoglobulin G (IgG) measurement unit (35130.1)	measurement unit used to record Immunoglobulin G (IgG)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i>
Immunoglobulin G (IgG) Upper Range (35131.1)	upper range of results for Immunoglobulin G (IgG)	0..1	testResult
Immunoglobulin G (IgG) Lower Range (35132.1)	upper range of results for Immunoglobulin G (IgG)	0..1	testResult

together with, in the case of

3.3.6.12.3 Immunoglobulin M (IgM) (35133.1)

Name	Description	Multiplicity	Data Type
Immunoglobulin M (IgM) result (35134.1)	Immunoglobulin M (IgM) result	1..1	testResult
Immunoglobulin M (IgM) measurement unit (35135.1)	measurement unit used to record Immunoglobulin M (IgM)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Immunoglobulin M (IgM) Upper Range (35136.1)	upper range of results for Immunoglobulin M (IgM)	0..1	testResult
Immunoglobulin M (IgM) Lower Range (35137.1)	upper range of results for Immunoglobulin M (IgM)	0..1	testResult

together with, in the case of

3.3.6.12.4 Immunoglobulin E (IgE) (35138.1)

Name	Description	Multiplicity	Data Type
Immunoglobulin E (IgE) result (35139.1)	Immunoglobulin E (IgE) result	1..1	testResult
Immunoglobulin E (IgE) measurement unit (35140.1)	measurement unit used to record Immunoglobulin E (IgE)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details

Immunoglobulin E (IgE) Upper Range (35141.1)	upper range of results for Immunoglobulin E (IgE)	0..1	testResult
Immunoglobulin E (IgE) Lower Range (35142.1)	upper range of results for Immunoglobulin E (IgE)	0..1	testResult

or, in the case of

3.3.6.13 CSF tests (30352.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34597.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeCSFtests <i>Enumeration</i> CSF tests: CSF tests
Specimen Type (34600.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType CSFtests <i>Enumeration</i> CSF: CSF
CSF amino acids (30349.2)	Please submit paired plasma result where available	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Neurotransmitters in CSF result (30350.2)	Neurotransmitters - Cerebrospinal fluid result	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition

			Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
B6/folate metabolites in CSF result (30351.2)	B6/folate metabolites - Cerebrospinal fluid result	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.13.1 Protein CSF (35143.1)

Name	Description	Multiplicity	Data Type
Protein CSF result (35144.1)	Protein CSF result	1..1	testResult
Protein CSF measurement unit (35145.1)	measurement unit used to record Protein CSF	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Protein CSF Upper Range (35146.1)	upper range of results for Protein CSF	0..1	testResult
Protein CSF Lower Range (35147.1)	upper range of results for Protein CSF	0..1	testResult

together with, in the case of

3.3.6.13.2 CSF lactate (35148.1)

Name	Description	Multiplicity	Data Type
CSF lactate result (35149.1)	CSF lactate result	1..1	testResult

CSF lactate measurement unit (35150.1)	measurement unit used to record CSF lactate	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
CSF lactate Upper Range (35151.1)	upper range of results for CSF lactate	0..1	testResult
CSF lactate Lower Range (35152.1)	upper range of results for CSF lactate	0..1	testResult

together with, in the case of

3.3.6.13.3 CSF glucose (35153.1)

Name	Description	Multiplicity	Data Type
CSF glucose result (35154.1)	CSF glucose result	1..1	testResult
CSF glucose measurement unit (35155.1)	measurement unit used to record CSF glucose	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
CSF glucose Upper Range (35156.1)	upper range of results for CSF glucose	0..1	testResult
CSF glucose Lower Range (35157.1)	upper range of results for CSF glucose	0..1	testResult

or, in the case of

3.3.6.14 Renal biochemistry (30355.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34603.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeRenalbiochemistry <i>Enumeration</i> Renal biochemistry: Renal biochemistry
Specimen Type (34606.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Renalbiochemistry <i>Enumeration</i> Blood: Blood

together with, in the case of

3.3.6.14.1 Serum Urea (35158.1)

Name	Description	Multiplicity	Data Type
Serum Urea result (35159.1)	Serum Urea result	1..1	testResult
Serum Urea measurement unit (35160.1)	measurement unit used to record Serum Urea	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> <i>>10 Enumerations please click above for more details</i>
Serum Urea Upper Range (35161.1)	upper range of results for Serum Urea	0..1	testResult
Serum Urea Lower Range (35162.1)	upper range of results for Serum Urea	0..1	testResult

together with, in the case of

3.3.6.14.2 Serum Creatinine (35163.1)

Serum Creatinine

Name	Description	Multiplicity	Data Type
Serum Creatinine result (35164.1)	Serum Creatinine result	1..1	testResult
Serum Creatinine measurement unit (35165.1)	measurement unit used to record Serum Creatinine	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> <i>>10 Enumerations please click above for more details</i>
Serum Creatinine Upper Range (35166.1)	upper range of results for Serum Creatinine	0..1	testResult
Serum Creatinine Lower Range (35167.1)	upper range of results for Serum Creatinine	0..1	testResult

together with, in the case of

3.3.6.14.3 Sodium

Name	Description	Multiplicity	Data Type
Sodium result (34859.1)	Sodium result	1..1	testResult

Sodium measurement unit (34860.1)	measurement unit used to record Sodium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Sodium Upper Range (34861.1)	upper range of results for Sodium	0..1	testResult
Sodium Lower Range (34862.1)	upper range of results for Sodium	0..1	testResult

together with, in the case of

3.3.6.14.4 Potassium

Name	Description	Multiplicity	Data Type
Potassium result (34869.1)	Potassium result	1..1	testResult
Potassium measurement unit (34870.1)	measurement unit used to record Potassium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Potassium Upper Range (34871.1)	upper range of results for Potassium	0..1	testResult
Potassium Lower Range (34872.1)	upper range of results for Potassium	0..1	testResult

or, in the case of

3.3.6.15 Blood Tests (30357.2)

Name	Description	Multiplicity	Data Type
Analytical technique (34609.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeBloodTests <i>Enumeration</i> Blood Tests: Blood Tests
Specimen Type (34612.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType BloodTests <i>Enumeration</i> Blood: Blood

together with, in the case of

3.3.6.15.1 Biotinidase (33142.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34615.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeBiotinidase <i>Enumeration</i> Biotinidase: Biotinidase

together with, in the case of

3.3.6.15.2 Bioitinidase (35168.1)

Name	Description	Multiplicity	Data Type
Bioitinidase result (35169.1)	Bioitinidase result	1..1	testResult
Bioitinidase measurement unit (35170.1)	measurement unit used to record Bioitinidase	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Bioitinidase Upper Range (35171.1)	upper range of results for Bioitinidase	0..1	testResult
Bioitinidase Lower Range (35172.1)	upper range of results for Bioitinidase	0..1	testResult

together with, in the case of

3.3.6.15.3 Clotting (33144.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34621.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeClotting <i>Enumeration</i> Clotting: Clotting
INR result (15569.2)	Clotting	0..1	xs:decimal

together with, in the case of

3.3.6.15.4 Coeliac antibodies (33145.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34627.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeCoeliacantibodies <i>Enumeration</i> Coeliac antibodies: Coeliac antibodies

Coeliac screen result (30387.2)	Coeliac screen	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Tissue transglutaminase antibody (33266.1)		0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Endomysial antibody (33267.1)		0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.15.5 Complement (33146.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34633.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeComplement <i>Enumeration</i> Complement: Complement
C3 Complement	Result of C3 complement test	0..1	C3 complement <i>Enumeration</i>

result (30803.1)			High: Raised above normal level Low: Reduced below normal level Normal: At a normal level
C4 complement result (30805.1)	Result of C4 complement test	0..1	C4 complement <i>Enumeration</i> High: Raised above normal level Low: Reduced below normal level Normal: At a normal level

together with, in the case of

3.3.6.15.6 Congenital Myasthenia Antibodies (33147.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34639.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeCongenitalMyastheniaAntibodies <i>Enumeration</i> Congenital Myasthenia Antibodies: Congenital Myasthenia Antibodies
Acetylcholine receptor antibodies (low affinity) result (28955.1)	Acetylcholine receptor antibodies (low affinity)	0..1	PositiveNegativeUnknown <i>Enumeration</i> unknown: unknown negative: negative positive: positive
Acetylcholine receptor antibodies (standard test) result (28933.1)	Acetylcholine receptor antibodies (standard test)	0..1	PositiveNegativeUnknown <i>Enumeration</i> unknown: unknown negative: negative positive: positive
MuSK antibodies result (28956.1)	MuSK antibodies	0..1	PositiveNegativeUnknown <i>Enumeration</i> unknown: unknown negative: negative positive: positive

together with, in the case of

3.3.6.15.7 Cortisol (33148.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34645.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeCortisol <i>Enumeration</i> Cortisol: Cortisol
Random cortisol result (28860.2)	Any abnormality in random cortisol level?	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.15.8 Cortisol (basal) (35173.1)

Name	Description	Multiplicity	Data Type
Cortisol (basal) result (35174.1)	Cortisol (basal) result	1..1	testResult
Cortisol (basal) measurement unit (35175.1)	measurement unit used to record Cortisol (basal)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Cortisol (basal) Upper Range (35176.1)	upper range of results for Cortisol (basal)	0..1	testResult
Cortisol (basal) Lower Range (35177.1)	upper range of results for Cortisol (basal)	0..1	testResult

together with, in the case of

3.3.6.15.9 Cortisol, peak stimulated (35178.1)

Name	Description	Multiplicity	Data Type
Cortisol, peak stimulated result (35179.1)	Cortisol, peak stimulated result	1..1	testResult
Cortisol, peak stimulated measurement unit (35180.1)	measurement unit used to record Cortisol, peak stimulated	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Cortisol, peak stimulated Upper Range (35181.1)	upper range of results for Cortisol, peak stimulated	0..1	testResult
Cortisol, peak stimulated Lower Range (35182.1)	upper range of results for Cortisol, peak stimulated	0..1	testResult

together with, in the case of

3.3.6.15.10 ACTH (35183.1)

Name	Description	Multiplicity	Data Type
ACTH result (35184.1)	ACTH result	1..1	testResult
ACTH measurement unit (35185.1)	measurement unit used to record ACTH	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
ACTH Upper Range (35186.1)	upper range of results for ACTH	0..1	testResult
ACTH Lower Range (35187.1)	upper range of results for ACTH	0..1	testResult

together with, in the case of

3.3.6.15.11 Extended haematology investigations (33151.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34657.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeExtendedhaematologyinvestigations <i>Enumeration</i> Extended haematology investigations: Extended haematology investigations

together with, in the case of

3.3.6.15.12 Haptoglobin (35188.1)

Name	Description	Multiplicity	Data Type
Haptoglobin result (35189.1)	Haptoglobin result	1..1	testResult
Haptoglobin measurement unit (35190.1)	measurement unit used to record Haptoglobin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Haptoglobin Upper Range (35191.1)	upper range of results for Haptoglobin	0..1	testResult
Haptoglobin Lower Range (35192.1)	upper range of results for Haptoglobin	0..1	testResult

together with, in the case of

3.3.6.15.13 Ferritin (35193.1)

Name	Description	Multiplicity	Data Type
Ferritin result (35194.1)	Ferritin result	1..1	testResult
Ferritin measurement unit (35195.1)	measurement unit used to record Ferritin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Ferritin Upper Range (35196.1)	upper range of results for Ferritin	0..1	testResult
Ferritin Lower Range (35197.1)	upper range of results for Ferritin	0..1	testResult

together with, in the case of

3.3.6.15.14 Extended renal biochemistry (33152.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34663.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeExtendedrenalbiochemistry <i>Enumeration</i> Extended renal biochemistry: Extended renal biochemistry

together with, in the case of

3.3.6.15.15 Creatinine (clearance) (35423.1)

Name	Description	Multiplicity	Data Type
Creatinine (clearance) result (35419.1)		1..1	xs:decimal
Creatinine (clearance) measurement unit (35420.1)		1..1	specialistRenalUnitOfMeasurement <i>Enumeration</i> Litres per week per 1.73 metres squared (l/week/1.73²): Litres per week per 1.73 metres squared (l/week/1.73 ²) Microgram albumin per 24 hours (µg/24hr): Microgram albumin per 24 hours (µg/24hr) Microgram albumin per hour (µg/ml/hr): Microgram albumin per hour (µg/ml/hr) Microgram albumin per minute (µg/min): Microgram albumin per minute (µg/min) Millilitres per Minute divided by 1.73 Square Metres (ml/min/1.73m²): Millilitres per Minute divided by 1.73 Square Metres (ml/min/1.73m ²)
Creatinine (clearance) Upper Range (35421.1)		0..1	xs:decimal
Creatinine (clearance) Lower Range (35422.1)		0..1	xs:decimal

together with, in the case of

3.3.6.15.16 eGFR (35428.1)

Name	Description	Multiplicity	Data Type
eGFR result (35424.1)	eGFR - Renal Biochemistry using MDRD equation	1..1	xs:decimal
eGFR measurement unit (35425.1)		1..1	specialistRenalUnitOfMeasurement <i>Enumeration</i> Litres per week per 1.73 metres squared (l/week/1.73²): Litres per week per 1.73 metres squared (l/week/1.73 ²)

			Microgram albumin per 24 hours (µg/24hr): Microgram albumin per 24 hours (µg/24hr) Microgram albumin per hour (µg/ml/hr): Microgram albumin per hour (µg/ml/hr) Microgram albumin per minute (µg/min): Microgram albumin per minute (µg/min) Millilitres per Minute divided by 1.73 Square Metres (ml/min/1.73m2): Millilitres per Minute divided by 1.73 Square Metres (ml/min/1.73m2)
eGFR Upper Range (35426.1)		0..1	xs:decimal
eGFR Lower Range (35427.1)		0..1	xs:decimal

together with, in the case of

3.3.6.15.17 Bicarbonate (35198.1)

Bicarbonate result

Name	Description	Multiplicity	Data Type
Bicarbonate result (35199.1)	Bicarbonate result	1..1	testResult
Bicarbonate measurement unit (35200.1)	measurement unit used to record Bicarbonate	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Bicarbonate Upper Range (35201.1)	upper range of results for Bicarbonate	0..1	testResult
Bicarbonate Lower Range (35202.1)	upper range of results for Bicarbonate	0..1	testResult

together with, in the case of

3.3.6.15.18 Magnesium (35203.1)

Name	Description	Multiplicity	Data Type
Magnesium result	Magnesium result	1..1	testResult

(35204.1)			
Magnesium measurement unit (35205.1)	measurement unit used to record Magnesium	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Magnesium Upper Range (35206.1)	upper range of results for Magnesium	0..1	testResult
Magnesium Lower Range (35207.1)	upper range of results for Magnesium	0..1	testResult

together with, in the case of

3.3.6.15.19 Oxalate (35208.1)

Name	Description	Multiplicity	Data Type
Oxalate result (35209.1)	Oxalate result	1..1	testResult
Oxalate measurement unit (35210.1)	measurement unit used to record Oxalate	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Oxalate Upper Range (35211.1)	upper range of results for Oxalate	0..1	testResult
Oxalate Lower Range (35212.1)	upper range of results for Oxalate	0..1	testResult

together with, in the case of

3.3.6.15.20 Chloride (35213.1)

Name	Description	Multiplicity	Data Type
Chloride result (35214.1)	Chloride result	1..1	testResult
Chloride measurement unit (35215.1)	measurement unit used to record Chloride	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Chloride Upper Range (35216.1)	upper range of results for Chloride	0..1	testResult
Chloride Lower Range (35217.1)	upper range of results for Chloride	0..1	testResult

together with, in the case of

3.3.6.15.21 Plasma osmolality (34878.1)

Name	Description	Multiplicity	Data Type
Plasma osmolality result (34879.1)	Plasma osmolality result	1..1	testResult
Plasma osmolality measurement unit (34880.1)		1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Plasma osmolality Upper Range (34881.1)	upper range of results for Plasma osmolality	0..1	testResult
Plasma osmolality Lower Range (34882.1)	upper range of results for Plasma osmolality	0..1	testResult

together with, in the case of

3.3.6.15.22 Glucose (33153.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34669.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeGlucose <i>Enumeration</i> Glucose: Glucose
Glucose Tolerance Test result (30399.2)	Glucose Tolerance Test	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.15.23 Fasting Glucose (35218.1)

Name	Description	Multiplicity	Data Type
Fasting Glucose result (35219.1)	Fasting Glucose result	1..1	testResult
Fasting Glucose measurement unit (35220.1)	measurement unit used to record Fasting Glucose	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Fasting Glucose Upper Range (35221.1)	upper range of results for Fasting Glucose	0..1	testResult
Fasting Glucose Lower Range (35222.1)	upper range of results for Fasting Glucose	0..1	testResult

together with, in the case of

3.3.6.15.24 Plasma Glucose (non-fasting) (35223.1)

Name	Description	Multiplicity	Data Type
Plasma Glucose (non-fasting) result (35224.1)	Plasma Glucose (non-fasting) result	1..1	testResult
Plasma Glucose (non-fasting) measurement unit (35225.1)	measurement unit used to record Plasma Glucose (non-fasting)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Plasma Glucose (non-fasting) Upper Range (35226.1)	upper range of results for Plasma Glucose (non-fasting)	0..1	testResult
Plasma Glucose (non-fasting) Lower Range (35227.1)	upper range of results for Plasma Glucose (non-fasting)	0..1	testResult

together with, in the case of

3.3.6.15.25 HbA1c (35228.1)

Name	Description	Multiplicity	Data Type
HbA1c result (35229.1)	HbA1c result	1..1	testResult

HbA1c measurement unit (35230.1)	measurement unit used to record HbA1c	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
HbA1c Upper Range (35231.1)	upper range of results for HbA1c	0..1	testResult
HbA1c Lower Range (35232.1)	upper range of results for HbA1c	0..1	testResult

together with, in the case of

3.3.6.15.26 Growth hormones (33154.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34675.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeGrowthhormones <i>Enumeration</i> Growth hormones: Growth hormones

3.3.6.15.27IGF1 (35233.1)

Name	Description	Multiplicity	Data Type
IGF1 result (35234.1)	IGF1 result	1..1	testResult
IGF1 measurement unit (35235.1)	measurement unit used to record IGF1	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
IGF1 Upper Range (35236.1)	upper range of results for IGF1	0..1	testResult
IGF1 Lower Range (35237.1)	upper range of results for IGF1	0..1	testResult

together with, in the case of

3.3.6.15.28 GFBP3 concentration (35238.1)

Name	Description	Multiplicity	Data Type
IGFBP3 concentration result (35239.1)	IGFBP3 concentration result	1..1	testResult
IGFBP3 concentration measurement unit	measurement unit used to record IGFBP3 concentration	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click

(35240.1)			<i>above for more details</i>
IGFBP3 concentration Upper Range (35241.1)	upper range of results for IGFBP3 concentration	0..1	testResult
IGFBP3 concentration Lower Range (35242.1)	upper range of results for IGFBP3 concentration	0..1	testResult

together with, in the case of

3.3.6.15.29 ALS (35243.1)

Name	Description	Multiplicity	Data Type
ALS result (35244.1)	ALS result	1..1	testResult
ALS measurement unit (35245.1)	measurement unit used to record ALS	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
ALS Upper Range (35246.1)	upper range of results for ALS	0..1	testResult
ALS Lower Range (35247.1)	upper range of results for ALS	0..1	testResult

together with, in the case of

3.3.6.15.30 Hormones (other) (33155.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34681.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeHormones(other) <i>Enumeration</i> Hormones (other): Hormones (other)

together with, in the case of

3.3.6.15.31 Prolactin (35248.1)

Name	Description	Multiplicity	Data Type
Prolactin result (35249.1)	Prolactin result	1..1	testResult
Prolactin	measurement unit used to	1..1	concentrationUnitOfMeasurement

measurement unit (35250.1)	record Prolactin		<i>Enumeration</i>
Prolactin Upper Range (35251.1)	upper range of results for Prolactin	0..1	testResult
Prolactin Lower Range (35252.1)	upper range of results for Prolactin	0..1	testResult

together with, in the case of

3.3.6.15.32 Inflammatory markers (33156.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34687.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeInflammatorymarkers <i>Enumeration</i> Inflammatory markers: Inflammatory markers

together with, in the case of

3.3.6.15.33 C reactive protein (35253.1)

Name	Description	Multiplicity	Data Type
C reactive protein result (35254.1)	C reactive protein result	1..1	testResult
C reactive protein measurement unit (35255.1)	measurement unit used to record C reactive protein	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
C reactive protein Upper Range (35256.1)	upper range of results for C reactive protein	0..1	testResult
C reactive protein Lower Range (35257.1)	upper range of results for C reactive protein	0..1	testResult

together with, in the case of

3.3.6.15.34 Cryoglobulins (33149.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34651.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeCryoglobulins <i>Enumeration</i> Cryoglobulins: Cryoglobulins

Cryoglobulin result (30393.2)	Cryoglobulin	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.15.35 Insulin and C-peptide (33157.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34693.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeInsulinandC-peptide <i>Enumeration</i> Insulin and C-peptide: Insulin and C-peptide
C-peptide result (28841.1)	C-peptide concentration in pmol/l	0..1	concentrationInPmol/l

together with, in the case of

3.3.6.15.36 Fasting Insulin (35258.1)

Name	Description	Multiplicity	Data Type
Fasting Insulin result (35259.1)	Fasting Insulin result	1..1	testResult
Fasting Insulin measurement unit (35260.1)	measurement unit used to record Fasting Insulin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Fasting Insulin Upper Range (35261.1)	upper range of results for Fasting Insulin	0..1	testResult

Fasting Insulin Lower Range (35262.1)	upper range of results for Fasting Insulin	0..1	testResult
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together with, in the case of

together with, in the case of

3.3.6.15.37 Random Insulin (35263.1)

Name	Description	Multiplicity	Data Type
Random Insulin result (35264.1)	Random Insulin result	1..1	testResult
Random Insulin measurement unit (35265.1)	measurement unit used to record Random Insulin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Random Insulin Upper Range (35266.1)	upper range of results for Random Insulin	0..1	testResult
Random Insulin Lower Range (35267.1)	upper range of results for Random Insulin	0..1	testResult

together with, in the case of

3.3.6.15.38 Lipids (33158.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34699.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeLipids <i>Enumeration</i> Lipids: Lipids

together with, in the case of

3.3.6.15.39 Total Cholesterol (Fasting) (35268.1)

Name	Description	Multiplicity	Data Type
Total Cholesterol (Fasting) result (35269.1)	Total Cholesterol (Fasting) result	1..1	testResult
Total Cholesterol (Fasting) measurement unit	measurement unit used to record Total Cholesterol	1..1	concentrationUnitOfMeasurement <i>Enumeration</i>

(35270.1)	(Fasting)		
Total Cholesterol (Fasting) Upper Range (35271.1)	upper range of results for Total Cholesterol (Fasting)	0..1	testResult
Total Cholesterol (Fasting) Lower Range (35272.1)	upper range of results for Total Cholesterol (Fasting)	0..1	testResult

together with, in the case of

3.3.6.15.40 Total Cholesterol (Non-fasting) (35273.1)

Name	Description	Multiplicity	Data Type
Total Cholesterol (Non-fasting) result (35274.1)	Total Cholesterol (Non-fasting) result	1..1	testResult
Total Cholesterol (Non-fasting) measurement unit (35275.1)	measurement unit used to record Total Cholesterol (Non-fasting)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Total Cholesterol (Non-fasting) Upper Range (35276.1)	upper range of results for Total Cholesterol (Non-fasting)	0..1	testResult
Total Cholesterol (Non-fasting) Lower Range (35277.1)	upper range of results for Total Cholesterol (Non-fasting)	0..1	testResult

together with, in the case of

3.3.6.15.41 Triglycerides (Non-fasting) (35278.1)

Triglycerides (Non-fasting) result

Name	Description	Multiplicity	Data Type
Triglycerides (Non-fasting) result (35279.1)	Triglycerides (Non-fasting) result	1..1	testResult
Triglycerides (Non-fasting) measurement unit (35280.1)	measurement unit used to record Triglycerides (Non-fasting)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Triglycerides (Non-fasting) Upper	upper range of results for Triglycerides (Non-fasting)	0..1	testResult

Range (35281.1)			
Triglycerides (Non-fasting) Lower Range (35282.1)	upper range of results for Triglycerides (Non-fasting)	0..1	testResult

together with, in the case of

3.3.6.15.42 Metabolic biochemistry (33160.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34705.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeMetabolicbiochemistry <i>Enumeration</i> Metabolic biochemistry: Metabolic biochemistry
Acylcarnitines result (30361.2)	Acylcarnitines	0..unbounded	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Amino acid result (30365.2)	Amino acid	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Plasma lysosomal enzymes (I-cell profile) result (30413.2)	Plasma lysosomal enzymes (I-cell profile)	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

White Cell Enzymes result (30433.2)	White Cell Enzymes	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Transferrin electrophoresis result (30429.2)	Transferrin electrophoresis	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Very long chain fatty acids (34188.1)	Very long chain fatty acid analysis	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.6.15.43 Ammonia (35283.1)

Name	Description	Multiplicity	Data Type
Ammonia result (35284.1)	Ammonia result	1..1	testResult
Ammonia measurement unit (35285.1)	measurement unit used to record Ammonia	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details

Ammonia Upper Range (35286.1)	upper range of results for Ammonia	0..1	testResult
Ammonia Lower Range (35287.1)	upper range of results for Ammonia	0..1	testResult

together with, in the case of

3.3.6.15.44 Lactate - Lab (35288.1)

Name	Description	Multiplicity	Data Type
Lactate - Lab result (35289.1)	Lactate - Lab result	1..1	testResult
Lactate - Lab measurement unit (35290.1)	measurement unit used to record Lactate - Lab	1..1	concentrationUnitOfMeasurement <i>Enumeration</i>
Lactate - Lab Upper Range (35291.1)	upper range of results for Lactate - Lab	0..1	testResult
Lactate - Lab Lower Range (35292.1)	upper range of results for Lactate - Lab	0..1	testResult

together with, in the case of

3.3.6.15.45 Plasma oxysterols (35293.1)

Name	Description	Multiplicity	Data Type
Plasma oxysterols result (35294.1)	Plasma oxysterols result	1..1	testResult
Plasma oxysterols measurement unit (35295.1)	measurement unit used to record Plasma oxysterols	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Plasma oxysterols Upper Range (35296.1)	upper range of results for Plasma oxysterols	0..1	testResult
Plasma oxysterols Lower Range (35297.1)	upper range of results for Plasma oxysterols	0..1	testResult

together with, in the case of

3.3.6.15.46 Blood copper (35298.1)

Name	Description	Multiplicity	Data Type
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Blood copper result (35299.1)	Blood copper result	1..1	testResult
Blood copper measurement unit (35300.1)	measurement unit used to record Blood copper	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Blood copper Upper Range (35301.1)	upper range of results for Blood copper	0..1	testResult
Blood copper Lower Range (35302.1)	upper range of results for Blood copper	0..1	testResult

together with, in the case of

3.3.6.15.47 Blood caeruloplasmin (35303.1)

Name	Description	Multiplicity	Data Type
Blood caeruloplasmin result (35304.1)	Blood caeruloplasmin result	1..1	testResult
Blood caeruloplasmin measurement unit (35305.1)	measurement unit used to record Blood caeruloplasmin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Blood caeruloplasmin Upper Range (35306.1)	upper range of results for Blood caeruloplasmin	0..1	testResult
Blood caeruloplasmin Lower Range (35307.1)	upper range of results for Blood caeruloplasmin	0..1	testResult

together with, in the case of

3.3.6.15.48 Uric acid (35308.1)

Name	Description	Multiplicity	Data Type
Uric acid result (35309.1)	Uric acid result	1..1	testResult
Uric acid measurement unit	measurement unit used to record Uric acid	1..1	concentrationUnitOfMeasurement <i>Enumeration</i>

(35310.1)			>10 Enumerations please click above for more details
Uric acid Upper Range (35311.1)	upper range of results for Uric acid	0..1	testResult
Uric acid Lower Range (35312.1)	upper range of results for Uric acid	0..1	testResult

together with, in the case of

3.3.6.15.49 Microbiology antibodies (33173.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34711.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeMicrobiologyantibodies <i>Enumeration</i> Microbiology antibodies: Microbiology antibodies
Aspergillus-specific IgG result (30371.2)	Aspergillus-specific IgG	0..1	antibodyResult <i>Enumeration</i> positive: positive negative: Negative unknown: Unknown significance not done: not done
Aspergillus-specific IgE result (30369.2)	Aspergillus-specific IgE	0..1	antibodyResult <i>Enumeration</i> positive: positive negative: Negative unknown: Unknown significance not done: not done

together with, in the case of

3.3.6.15.50 Porphyrin investigations (33164.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34717.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypePorphyrininvestigations <i>Enumeration</i> Porphyrin investigations: Porphyrin investigations

together with, in the case of

3.3.6.15.51 Ferrochelatase enzyme activity (normal) (35313.1)

Name	Description	Multiplicity	Data Type
Ferrochelatase enzyme activity (normal) result (35314.1)	Ferrochelatase enzyme activity (normal) result	1..1	testResult
Ferrochelatase enzyme activity (normal) measurement unit (35315.1)	measurement unit used to record Ferrochelatase enzyme activity (normal)	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Ferrochelatase enzyme activity (normal) Upper Range (35316.1)	upper range of results for Ferrochelatase enzyme activity (normal)	0..1	testResult
Ferrochelatase enzyme activity (normal) Lower Range (35317.1)	upper range of results for Ferrochelatase enzyme activity (normal)	0..1	testResult

together with, in the case of

3.3.6.15.52 Primary immunodeficiency investigations (33165.1).

Name	Description	Multiplicity	Data Type
Analytical technique (34723.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypePrimaryimmunodeficiencyinvestigations <i>Enumeration</i> Primary immunodeficiency investigations: Primary immunodeficiency investigations
CD19+ B level result (28872.1)	CD19+ B levels	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
CD3 proliferation test result (28873.1)	CD3 proliferation tests	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High

CD3+ CD4+ T Helper levels result (28869.1)	CD3+ CD4+ T Helper levels	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
CD3+ CD8+ Cytotoxic T levels result (28870.1)	CD3+ CD8+ Cytotoxic T levels	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
CD56+ NK levels result (28871.1)	CD56+ NK levels	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
IL12/IFN response result (28875.1)	IL12/IFN response	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
Neutrophil response result (28876.1)	Neutrophil response	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
TLR signalling result (28874.1)	TLR signalling	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High

together with, in the case of

3.3.6.15.53 Renin and aldosterone (33166.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34729.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeReninandaldosterone <i>Enumeration</i> Renin and aldosterone: Renin and aldosterone

together with, in the case of

3.3.6.15.54 Aldosterone (35318.1)

Name	Description	Multiplicity	Data Type
Aldosterone result (35319.1)	Aldosterone result	1..1	testResult
Aldosterone measurement unit (35320.1)	measurement unit used to record Aldosterone	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Aldosterone Upper Range (35321.1)	upper range of results for Aldosterone	0..1	testResult
Aldosterone Lower Range (35322.1)	upper range of results for Aldosterone	0..1	testResult

together with, in the case of

3.3.6.15.55 Renin (35323.1)

Name	Description	Multiplicity	Data Type
Renin result (35324.1)	Renin result	1..1	testResult
Renin measurement unit (35325.1)	measurement unit used to record Renin	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Renin Upper Range (35326.1)	upper range of results for Renin	0..1	testResult
Renin Lower Range (35327.1)	upper range of results for Renin	0..1	testResult

together with, in the case of

3.3.6.15.56 Renin:aldosterone ratio (35328.1)

Name	Description	Multiplicity	Data Type
Renin:aldosterone ratio result (35329.1)	Renin:aldosterone ratio result	1..1	testResult
Renin:aldosterone ratio Upper Range (35331.1)	upper range of results for Renin:aldosterone ratio	0..1	testResult
Renin:aldosterone ratio Lower Range (35332.1)	upper range of results for Renin:aldosterone ratio	0..1	testResult

together with, in the case of

3.3.6.15.57 Renin activity (35333.1)

Name	Description	Multiplicity	Data Type
Renin activity result (35334.1)	Renin activity result	1..1	testResult
Renin activity measurement unit (35335.1)	measurement unit used to record Renin activity	1..1	concentrationUnitOfMeasurement <i>Enumeration</i> >10 Enumerations please click above for more details
Renin activity Upper Range (35336.1)	upper range of results for Renin activity	0..1	testResult
Renin activity Lower Range (35337.1)	upper range of results for Renin activity	0..1	testResult

together with, in the case of

3.3.6.15.58 Sex hormones (33167.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34735.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeSexhormones <i>Enumeration</i> Sex hormones: Sex hormones

together with, in the case of

3.3.6.15.59 17-hydroxyprogesterone (35338.1)

Name	Description	Multiplicity	Data Type
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17-hydroxyprogesterone result (35339.1)	17-hydroxyprogesterone result	1..1	testResult
17-hydroxyprogesterone measurement unit (35340.1)	measurement unit used to record 17-hydroxyprogesterone	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
17-hydroxyprogesterone Upper Range (35341.1)	upper range of results for 17-hydroxyprogesterone	0..1	testResult
17-hydroxyprogesterone Lower Range (35342.1)	upper range of results for 17-hydroxyprogesterone	0..1	testResult

together with, in the case of

3.3.6.15.60 Sex steroid (35343.1)

Name	Description	Multiplicity	Data Type
Sex steroid result (35344.1)	Sex steroid result	1..1	testResult
Sex steroid measurement unit (35345.1)	measurement unit used to record Sex steroid	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
Sex steroid Upper Range (35346.1)	upper range of results for Sex steroid	0..1	testResult
Sex steroid Lower Range (35347.1)	upper range of results for Sex steroid	0..1	testResult

together with, in the case of

3.3.6.15.61 FSH (35348.1)

Name	Description	Multiplicity	Data Type
FSH result (35349.1)	FSH result	1..1	testResult
FSH measurement unit (35350.1)	measurement unit used to record FSH	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
FSH Upper Range	upper range of results for FSH	0..1	testResult

(35351.1)			
FSH Lower Range (35352.1)	upper range of results for FSH	0..1	testResult

together with, in the case of

3.3.6.15.62 hCG (35353.1)

Name	Description	Multiplicity	Data Type
hCG result (35354.1)	hCG result	1..1	testResult
hCG measurement unit (35355.1)	measurement unit used to record hCG	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
hCG Upper Range (35356.1)	upper range of results for hCG	0..1	testResult
hCG Lower Range (35357.1)	upper range of results for hCG	0..1	testResult

together with, in the case of

3.3.6.15.63 LH (35358.1)

Name	Description	Multiplicity	Data Type
LH result (35359.1)	LH result	1..1	testResult
LH measurement unit (35360.1)	measurement unit used to record LH	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
LH Upper Range (35361.1)	upper range of results for LH	0..1	testResult
LH Lower Range (35362.1)	upper range of results for LH	0..1	testResult

together with, in the case of

3.3.6.15.64 Thyroid function testing (33168.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34741.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeThyroidfunctiontesting Enumeration Thyroid function testing:Thyroid

			function testing
TSH result (33296.1)	Thyroid stimulating hormone	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
T3 result (33297.1)	T3 result	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
T4 result (33298.1)	T4 result	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
Free T4 result (33300.1)	Free T4 result	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High
Free T3 result (33299.1)	Free T3 result	0..1	LowNormalHighNotRecorded <i>Enumeration</i> Not Recorded: Not Recorded Low: Low Normal: Normal High: High

together with, in the case of

3.3.6.15.65 Virology (33171.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34747.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeVirology <i>Enumeration</i> Virology: Virology

Hepatitis B surface antigen (qual) result (15535.2)	Hepatitis B surface antigen (qual)	0..1	antibodyResult <i>Enumeration</i> positive :positive negative :Negative unknown :Unknown significance not done :not done
Hepatitis C antigen result (30405.2)	Hepatitis C antigen	0..1	antibodyResult <i>Enumeration</i> positive :positive negative :Negative unknown :Unknown significance not done :not done

together with, in the case of

3.3.6.15.66 EBV viral load (35363.1)

Name	Description	Multiplicity	Data Type
EBV viral load result (35364.1)	EBV viral load result	1..1	testResult
EBV viral load measurement unit (35365.1)	measurement unit used to record EBV viral load	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
EBV viral load Upper Range (35366.1)	upper range of results for EBV viral load	0..1	testResult
EBV viral load Lower Range (35367.1)	upper range of results for EBV viral load	0..1	testResult

together with, in the case of

3.3.6.15.67 Vitamin B12 (33172.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34753.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeVitaminB12 <i>Enumeration</i> Vitamin B12 :Vitamin B12

together with, in the case of

3.3.6.15.68 Vitamin B12 (35368.1)

Name	Description	Multiplicity	Data Type
Vitamin B12 result (35369.1)	Vitamin B12 result	1..1	testResult
Vitamin B12 measurement unit (35370.1)	measurement unit used to record Vitamin B12	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
Vitamin B12 Upper Range (35371.1)	upper range of results for Vitamin B12	0..1	testResult
Vitamin B12 Lower Range (35372.1)	upper range of results for Vitamin B12	0..1	testResult

together with, in the case of

3.3.6.15.69 Folate (35373.1)

Name	Description	Multiplicity	Data Type
Folate result (35374.1)	Folate result	1..1	testResult
Folate measurement unit (35375.1)	measurement unit used to record Folate	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
Folate Upper Range (35376.1)	upper range of results for Folate	0..1	testResult
Folate Lower Range (35377.1)	upper range of results for Folate	0..1	testResult

Or, in the case of

3.3.6.16 Other enzymes (33123.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34759.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeOtherenzymes Enumeration Other enzymes: Other enzymes
Specimen Type	Indicated the specimen type	1..1	analysedSpecimenType

(34762.1)	used for analysis		Otherenzymes <i>Enumeration</i> Blood: Blood
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together with, in the case of

3.3.6.16.1 Creatine kinase (35378.1)

Name	Description	Multiplicity	Data Type
Creatine kinase result (35379.1)	Creatine kinase result	1..1	testResult
Creatine kinase measurement unit (35380.1)	measurement unit used to record Creatine kinase	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
Creatine kinase Upper Range (35381.1)	upper range of results for Creatine kinase	0..1	testResult
Creatine kinase Lower Range (35382.1)	upper range of results for Creatine kinase	0..1	testResult

together with, in the case of

3.3.6.16.2 LDH (35383.1)

Name	Description	Multiplicity	Data Type
LDH result (35384.1)	LDH result	1..1	testResult
LDH measurement unit (35385.1)	measurement unit used to record LDH	1..1	concentrationUnitOfMeasurement >10 Enumerations please click above for more details
LDH Upper Range (35386.1)	upper range of results for LDH	0..1	testResult
LDH Lower Range (35387.1)	upper range of results for LDH	0..1	testResult

together with, in the case of

3.3.6.16.3 Aldolase (35388.1)

Name	Description	Multiplicity	Data Type
Aldolase result	Aldolase result	1..1	testResult

(35389.1)			
Aldolase measurement unit (35390.1)	measurement unit used to record Aldolase	1..1	concentrationUnitOfMeasurement <i>Enumeration</i>
Aldolase Upper Range (35391.1)	upper range of results for Aldolase	0..1	testResult
Aldolase Lower Range (35392.1)	upper range of results for Aldolase	0..1	testResult

or, in the case of

3.3.6.17 Fecal test (33308.1)

Name	Description	Multiplicity	Data Type
Analytical technique (34765.1)	If relevant please state physical analytic technique or protocol used for the test.	1..1	labAnalysisTypeFecaltest <i>Enumeration</i> Fecal test: Fecal test
Specimen Type (34768.1)	Indicated the specimen type used for analysis	1..1	analysedSpecimenType Fecaltest <i>Enumeration</i> Faeces: Faeces
Fecal fat result (33310.1)	Fecal fat result	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Fecal elastase result (33309.1)	Fecal elastase result	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the

			<p>patient's condition</p> <p>Abnormal-Unknown</p> <p>Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>
<p>Fecal trypsin result (33311.1)</p>	<p>Fecal trypsin result</p>	<p>0..1</p>	<p>Clinical Test Abnormality Enumeration</p> <p>Normal:Normal</p> <p>Unknown:No results available</p> <p>Abnormal-Relevant:An abnormality of clinical relevance to the patient's condition</p> <p>Abnormal-Unknown</p> <p>Relevance:An abnormality of unknown clinical relevance to the patient's condition</p>

3.3.7 Non-imaging Diagnostics (30107.2)

All non-imaging diagnostic reports should include the following elements,

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique other (30459.1)	Other non-imaging analytic technique - if not in list supplied	0..1	xs:string
Assessment (29524.1)	Assessment of findings and clinical significance	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Patient Status (34004.1)	Represents the patient's status when test was performed	1..1	patientStatus <i>Enumeration</i> presenting: Taken at patient presentation diagnostic: Taken at the point of diagnosis baseline: Representing a baseline measurement most abnormal: Representing the most abnormal measurement unknown: Status unknown
Report (29501.2)	File name of uploaded copy of clinical report - requested format [Participant ID]_[Local Report Identifier]	0..1	xs:base64Binary

together with, in the case of

General Non-imaging Diagnostics (34838.1)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (30178.2)	Non-imaging technique for analysis - chosen from list supplied	1..1	non-imagingMethods >10 Enumerations please click above for more details

together with, in the case of

3.3.7.1 Heart observations (30179.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34771.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodHeartobservations Enumeration Heart observations: Heart observations
Heart rate (28040.1)	Heart rate (ECG) measurement	1..1	xs:integer
Heart rhythm (28043.1)	Heart rhythm	0..1	heartRhythm Enumeration

or, in the case of

3.3.7.2 Forced vital capacity (30180.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34774.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodForcedvitalcapacity Enumeration Forced vital capacity: Forced vital capacity
Forced vital	Forced vital capacity/expected	1..1	FVCpercentage

capacity/expected forced vital capacity result (30181.1)	forced vital capacity result		
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or, in the case of

3.3.7.3 ECG diagnostics (30183.2)

Cardiac measurements made using electrocardiography.

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34777.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodECGdiagnostics <i>Enumeration</i> ECG diagnostics :ECG diagnostics
Type 1 ECG (31392.1)	Is a type 1 ECG present?	0..1	type1ECG <i>Enumeration</i> yes :Yes no :No unknown :Unknown
High RV leads (31394.1)	Are high RV leads recorded?	0..1	highRVleads <i>Enumeration</i> yes :Yes no :No unknown :Unknown
High RV in which lead? (31397.1)	Which lead has high RV?	0..unbounded	xs:string
QT interval (28600.1)	QT interval in ms	0..1	timeInMs
PR interval (28601.1)	Duration of PR interval in ms	0..1	timeInMs
QRS duration (28602.1)	Duration of QRS in ms	0..1	timeInMs
QRS axis (28605.1)	QRS axis in degrees	0..1	Axis
Pre-excitation (28617.2)	Does the ECG show any evidence of pre-excitation	0..1	yesNoUnk <i>Enumeration</i> yes :Yes no :No unknown :Unknown
Abnormal Q waves (28618.2)	Does the ECG show any abnormal Q waves?	0..1	yesNoUnk <i>Enumeration</i>

			yes: Yes no: No unknown: Unknown
Interventricular delay (31396.1)	Is there interventricular delay?	0..1	interventricularDelay <i>Enumeration</i> yes: Yes no: No unknown: Unknown

or, in the case of

3.3.7.4 Sleep test (30184.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34780.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodSleeptest <i>Enumeration</i> Sleep test: Sleep test
Mean sleep latency result (30185.1)	Mean sleep latency	0..1	xs:string
Number of SOREM (30186.1)	Sleep Onset Rapid Eye Movement number	0..1	xs:decimal
Total Sleep Time (30187.1)	Total sleep time in hours	0..1	xs:decimal
Sleep Related Breathing Parameters (30190.1)	Sleep Related Breathing Parameters	0..1	xs:string
Sleep Efficiency (30188.1)	Sleep Efficiency	0..1	xs:string
PLMS (30189.1)	Periodic limb movements in sleep	0..1	xs:string

or, in the case of

3.3.7.5 Cardiac Drug Challenge (31402.2)

Name	Description	Multiplicity	Data Type
Patient Status (31400.2)	Describes the status of a patient when ECG was taken	0..unbounded	patientStatus <i>Enumeration</i> presenting: Taken at

			patient presentation diagnostic :Taken at the point of diagnosis baseline :Representing a baseline measurement most abnormal :Representing the most abnormal measurement unknown :Status unknown
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together with, in the case of

3.3.7.5.1 Sodium channel blocker challenge (31403.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34786.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodSodiumchannelblockerchallenge <i>Enumeration</i> Sodium channel blocker challenge :Sodium channel blocker challenge
Sodium channel blocker (31407.1)	The sodium channel blocker used in a drug challenge test	1..1	sodiumChannelBlocker <i>Enumeration</i> ajmanline :Ajmanline flecainide :Flecainide
Sodium channel blocker dosage (31409.1)	The dosage of sodium channel blocker used in a cardiac drug challenge test	1..1	sodiumChannelBlockerDosage
Body weight at drug challenge (31410.1)	The person's body weight at the time of the drug challenge	1..1	personObservation(weight)
Calculated drug amount (31474.1)	Calculated amount of drug for a cardiac drug challenge test	0..1	calculatedDrugAmount
Drug dose at which diagnostic type 1 ECG appeared (31412.1)	Drug dose at which diagnostic type 1 ECG appeared	0..1	drugDose

Total amount of drug given (31413.1)	Total amount of drug given in drug challenge test	1..1	drugDose
Premature termination (31421.1)	Premature termination of a drug challenge test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Reason for premature termination (31415.1)	Reason for premature termination before a standard maximum dose was infused in a drug challenge test	0..1	xs:string
High RV leads used? (31416.1)	Were high RV leads used in a drug challenge test?	1..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Lead of first observation (31417.1)	In which lead was type 1 BrS ECG first observed?	0..1	xs:string

together with, in the case of

3.3.7.5.2 Epinephrine challenge (31404.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34789.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodEpinephrinechallenge <i>Enumeration</i> Epinephrine challenge: Epinephrine challenge
Epinephrine protocol (31420.1)	The epinephrine cardiac drug challenge protocol used	0..1	epinephrineProtocol <i>Enumeration</i> Shimizu: Shimizu Mayo clinic: Mayo clinic Other: Other
Premature termination	Premature termination of a drug challenge test	0..1	yesNoUnk <i>Enumeration</i> yes: yes

(31421.1)			no: no unknown: unknown
Reason for premature termination (31415.1)	Reason for premature termination before a standard maximum dose was infused in a drug challenge test	0..1	xs:string
Diagnostic QT prolongation (31422.1)	Diagnostic QT prolongation in cardiac drug challenge test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Diagnostic polymorphic VT (31423.1)	Diagnostic polymorphic VT	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Diagnostic bidirectional VT (31446.1)	Diagnostic bidirectional VT	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Diagnostic VPB burden (31424.1)	Diagnostic VPB burden in cardiac drug challenge test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Diagnostic VPB frequency (31425.1)	Description of VPB frequency	0..1	xs:string

or, in the case of

3.3.7.6 Holter monitor test (31432.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34792.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodHoltermonitortest <i>Enumeration</i> Holter monitor test: Holter monitor test

VPB burden/24 hours (31433.1)	VPB per 24 hours	0..1	xs:integer
Dynamic type 1 ECG pattern (31434.1)	Dynamic type 1 ECG pattern present	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
QT abnormalities detected (31435.1)	Were QT abnormalities detected in Holter monitor test	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
QT abnormality description (31436.1)	Description of QT abnormalities in Holter monitor test	0..1	xs:string
Holter Monitor Abnormalities (31462.1)	Abnormalities present on the Holter Monitor	0..1	holterMonitor <i>Enumeration</i> Atrial fibrillation: Atrial fibrillation Non-sustained VT: Non-sustained VT Nil: Nil Unknown: Unknown

or, in the case of

3.3.7.7 Signal averaged ECG (31437.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34795.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodSignalaveragedECG <i>Enumeration</i> Signal averaged ECG: Signal averaged ECG
fQRSd (31442.1)	fQRSd measurement from signal averaged ECG	0..1	fQRSd
HFLA (31443.1)	HFLA measurement from signal averaged ECG	0..1	HFLA
RMS (31444.1)	RMS measurment from signal averaged ECG	0..1	RMS

or, in the case of

3.3.7.8 Exercise test - cardiac (31445.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34798.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodExercisetest-cardiac <i>Enumeration</i> Exercise test - cardiac: Exercise test - cardiac
Diagnostic polymorphic VT (31423.1)	Diagnostic polymorphic VT	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Diagnostic bidirectional VT (31446.1)	Diagnostic bidirectional VT	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Dynamic type 1 ECG pattern (31434.1)	Dynamic type 1 ECG pattern present	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Recovery QTc>480ms (31447.1)	Recovery QTc interval greater than 480ms	0..1	yesNoUnk <i>Enumeration</i> yes: yes no: no unknown: unknown
Exercise test additional findings (31448.1)	Any additional findings from exercise test	0..unbounded	xs:string
Vasoactive medications (31463.1)	List any vasoactive medications that the patient was on at the time of an exercise tolerance test	0..unbounded	xs:string

or, in the case of

3.3.7.9 Electrophysiological study (31449.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34801.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodElectrophysiologicalstudy <i>Enumeration</i> Electrophysiological study: Electrophysiological study
VERP (31451.1)	VERP at S1 drive train 600ms	0..1	verp
Inducibility (31452.1)	Inducibility in EP study	0..1	yesNoUnk <i>Enumeration</i> yes: Yes no: No unknown: Unknown
Inducibility intervals (31453.1)	Specify inducibility intervals in EP study	0..1	xs:string

or, in the case of

3.3.7.10 Auditory Brainstem Response (32992.1)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34804.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodAuditoryBrainstemResponse <i>Enumeration</i> Auditory Brainstem Response: Auditory Brainstem Response
Auditory Brainstem Response (32999.1)	Result of an Auditory Brainstem Response (ABR) test	0..1	auditoryBrainstemResponseResult <i>Enumeration</i> Normal: Normal Abnormal: Abnormal Abnormal with no evidence of peaks: Abnormal with no evidence of peaks Abnormal with early evidence of peaks (up to wave III): Abnormal with early evidence of peaks (up to wave III) Abnormal morphology but wave V present: Abnormal morphology but wave V present

or, in the case of

3.3.7.11 Otoacoustic Emissions (32993.1)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34807.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodOtoacousticEmissions <i>Enumeration</i> Otoacoustic Emissions: Otoacoustic Emissions
Otoacoustic Emissions (32998.1)	Result of Otoacoustic Emissions (OAE) test	0..1	otoacousticEmissionsResult <i>Enumeration</i> Present: Present Absent: Absent Disappeared but Cochlear microphonic still present: Disappeared but Cochlear microphonic still present Disappeared and cochlear microphonic absent: Disappeared and cochlear microphonic absent

or, in the case of

3.3.7.12 Ocular metrics (30110.2)

3.3.7.12.1 Ocular Malformation Metrics (30260.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34813.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodOcularMalformationMetrics <i>Enumeration</i> Ocular Malformation Metrics: Ocular Malformation Metrics
Axial length - OD (28716.1)	Axial length in mm - Right Eye	0..1	lengthInMm
Axial length - OS (28717.1)	Axial length in mm - Left Eye	0..1	lengthInMm
Central corneal thickness - OD (28731.1)	Central corneal thickness in micrometres - Right Eye	0..1	thicknessInMicrometres
Central corneal thickness - OS (28732.1)	Central corneal thickness in micrometres - Left Eye	0..1	thicknessInMicrometres
Corneal diameter (horizontal) - OD (28723.1)	Corneal Diameter in mm - Right Eye	0..1	lengthInMm

Corneal diameter (horizontal) - OS (28724.1)	Corneal Diameter in mm - Left Eye	0..1	lengthInMm
Keratometry - OD (28721.1)	Corneal Curvature in dioptres - Right Eye	0..1	cornealCurvature
Keratometry - OS (28722.1)	Corneal Curvature in dioptres - Left Eye	0..1	cornealCurvature
Gonioscopy result (33077.1)	Results of gonioscopy	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

or, in the case of

3.3.7.12.2 Visual Field (30264.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34816.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	0..unbounded	nonImagingMethodVisualField <i>Enumeration</i> Visual Field: Visual Field
Central visual field (III, HVF) result (30262.1)	Humphrey Visual Field test result	0..1	degreesAngle
Continuous central visual field (III.4.e, GVF) (30263.1)	Goldmann visual field result using Goldmann isopter III-4e	0..1	degreesAngle

or, in the case of

3.3.7.12.3 Ocular Pressure (30265.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34819.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodOcularPressure <i>Enumeration</i> Ocular Pressure: Ocular Pressure
Intraocular pressure - OD (28713.1)	Intraocular pressure - Right eye	0..1	intraocularPressure

Intraocular pressure - OS (28714.1)	Intraocular pressure - Left eye	0..1	intraocularPressure
Intraocular Pressure - OD (highest recorded) (30266.1)	Highest recorded intraocular pressure in the right eye	0..1	intraocularPressure
Intraocular pressure - OS (highest recorded) (30267.1)	Highest recorded intraocular pressure in the left eye	0..1	intraocularPressure

or, in the case of

3.3.7.12.4 Colour Plate Test (30269.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34822.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodColourPlateTest <i>Enumeration</i> Colour Plate Test: Colour Plate Test
Ishihara colour plate result (30268.1)	Result of Ishihara color blindness test	1..1	PassFail <i>Enumeration</i> Fail: Fail Pass: Pass

or, in the case of

3.3.7.12.5 Electro-oculogram (30271.2).

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34825.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodElectro-oculogram <i>Enumeration</i> Electro-oculogram: Electro-oculogram
Arden ratio result (30270.1)	Arden ratio result from electro-oculogram test	1..1	xs:double

or, in the case of

3.3.7.12.6 Dark Adaptation Test (30276.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical	If relevant please state non-imaging analytic technique or	1..1	nonImagingMethodDarkAdaptationTest <i>Enumeration</i>

technique (34828.1)	protocol used for the test.		Dark Adaptation Test: Dark Adaptation Test
Dark adaptation final threshold result (30275.2)	Dark adaptation final threshold measurement	1..1	normalElevated <i>Enumeration</i> Normal: normal Elevated: elevated Unknown: unknown

or, in the case of

or, in the case of

3.3.7.12.7 Electroretinogram (30291.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34831.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodElectroretinogram <i>Enumeration</i> Electroretinogram: Electroretinogram
A-wave implicit time - OS (30281.1)	A-wave implicit time in left eye	0..1	timeIntervalMs
A-wave implicit time - OD (30282.1)	A-wave implicit time in right eye	0..1	timeIntervalMs
A-wave amplitude - OS (30279.1)	A-wave amplitude in left eye	0..1	amplitude
A-wave amplitude - OD (30280.1)	A-wave amplitude in right eye	0..1	amplitude
B-wave implicit time - OD (30286.1)	B-wave implicit time in right eye	0..1	timeIntervalMs
B-wave implicit time - OS (30287.1)	B-wave implicit time in left eye	0..1	timeIntervalMs
B-wave amplitude - OS (30283.1)	B-wave amplitude in left eye	0..1	amplitude
B-wave amplitude - OD (30284.1)	B-wave amplitude in right eye	0..1	amplitude
Electroretinogram response (30290.1)	Electroretinogram response	0..1	electroretinogramResponse <i>Enumeration</i> reduced: Reduced extinguished: Extinguished

Adaptation (30296.2)	Adaptation used in electroretinogram test	1..1	adaptation <i>Enumeration</i> dark :Dark adaptation used in ERG light :Light adaptation used in ERG
Stimulation (30297.1)	Stimulation used in electroretinogram test	1..1	stimulation <i>Enumeration</i> flicker :Flicker stimulation mixed/combined :Mixed/Combined stimulation

or, in the case of

3.3.7.12.8 Visual Acuity (30970.2)

Name	Description	Multiplicity	Data Type
Non-imaging analytical technique (34834.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodVisualAcuity <i>Enumeration</i> Visual Acuity :Visual Acuity
Visual acuity - OD (28734.1)	Visual acuity - Right Eye	0..1	visualAcuity <i>Enumeration</i> 20/_ :20/_ CF :CF HM :HM LP :LP NLP :NLP
Visual acuity - OS (28735.1)	Visual acuity - Left Eye	0..1	visualAcuity <i>Enumeration</i> 20/_ :20/_ CF :CF HM :HM LP :LP NLP :NLP

or, in the case of

3.3.7.12.9 Refraction Error (30978.2)

Name	Description	Multiplicity	Data Type
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Non-imaging analytical technique (34837.1)	If relevant please state non-imaging analytic technique or protocol used for the test.	1..1	nonImagingMethodRefractionError <i>Enumeration</i> Refraction Error: Refraction Error
Refraction Error (30977.1)	Measurement of the refraction error of the eye	0..1	xs:decimal

3.3.8 Patient History (30108.2)

All patient history reports should include the following elements,

Name	Description	Multiplicity	Data Type
Assessment (29524.1)	Assessment of findings and clinical significance	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
OMIM Code (29827.1)	OMIM code that best describes disorder	0..1	xs:string
Report (29501.2)	File name of uploaded copy of clinical report - requested format [Participant ID]_[Local Report Identifier]	0..1	xs:base64Binary

together with, in the case of

3.3.8.1 Age at transplant loss (30197.2)

Name	Description	Multiplicity	Data Type
Age at transplant loss (30192.2)	Age in years that transplant loss occurred	1..1	ageOfOnsetYears

or, in the case of

3.3.8.2 Age at diagnosis of chronic kidney disease (30198.2)

Name	Description	Multiplicity	Data Type
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Age at diagnosis of chronic kidney disease (30193.2)	Age of onset in years of chronic kidney disease	1..1	ageOfOnsetYears
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or, in the case of

3.3.8.3 Age at ESRD (30199.2)

Name	Description	Multiplicity	Data Type
Age at ESRD (30194.2)	Age of onset in years of End Stage Renal Disease	1..1	ageOfOnsetYears

or, in the case of

3.3.8.4 Inflammation metrics (30200.2)

Name	Description	Multiplicity	Data Type
Age of onset inflammatory symptoms (30195.2)	Age in years of onset of inflammatory symptoms	1..1	ageOfOnsetYears
Duration of inflammation attacks (33320.1)	Duration of inflammation attacks	0..1	durationOfInflammation

or, in the case of

3.3.8.5 Age of onset of motor symptoms (30201.2)

Name	Description	Multiplicity	Data Type
Age at onset of motor symptoms (28939.2)	Age at onset of motor symptoms	1..1	ageOfOnsetYears

or, in the case of

or, in the case of

3.3.8.6 Infection metrics (30202.1)

Name	Description	Multiplicity	Data Type
Infection frequency over the past 3 years (28879.1)	Typical infection frequency over the past 3 years	1..1	infectionFrequency <i>Enumeration</i> 0-1 per year: 0-1 per year 2-4 per year: 2-4 per year >4 per year: >4 per year Unknown: Unknown No Response: No Response
Typical infection duration over the past 3 years (28882.1)	Typical infection duration over the past 3 years	1..1	infectionDuration <i>Enumeration</i> None: None <7 days: <7 days 7-13 days: 7-13 days 14-42 days: 14-42 days >42 days: >42 days Unknown: Unknown No Response: No Response

or, in the case of

3.3.8.7 APGAR score (30207.1)

Name	Description	Multiplicity	Data Type
APGAR score (1 minute) result (28660.1)	APGAR score at 1 minute	0..1	apgarScore
APGAR score (5 minutes) result (28661.1)	APGAR score at 5 minutes	0..1	apgarScore

or, in the case of

3.3.8.8 Lifestyle factors (31470.2)

together with, in the case of

3.3.8.8.1 Exercise status (31469.1)

Name	Description	Multiplicity	Data Type
Exercise status	The level of exercise for the patient	0..1	exerciseStatus

(31468.1)			<i>Enumeration</i> Low: Low Moderate: Moderate Intense: Intense
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together with, in the case of

3.3.8.8.2 Alcohol intake (30206.1)

Name	Description	Multiplicity	Data Type
Alcohol Consumption (14447.1)	The ALCOHOL WEEKLY UNITS reported by the patient.	1..1	xs:integer

together with, in the case of

3.3.8.8.3 Smoking status (31464.1)

Name	Description	Multiplicity	Data Type
SMOKING STATUS (13980.1)	Specify the current smoking status of the patient.	0..1	smokingStatus <i>Enumeration</i> 1: Current smoker 2: Ex smoker 3: Non-smoker - history unknown 4: Never smoked Z: Not Stated (PERSON asked but declined to provide a response) 9: Unknown

or, in the case of

3.3.8.9 Renal history (33315.1)

Name	Description	Multiplicity	Data Type
Renal trigger (33314.1)	Known or suspected renal triggers	0..1	xs:string

3.3.9 Performance Measures (30109.2)

All performance measures reports should include the following elements,

Name	Description	Multiplicity	Data Type
Assessment (29524.1)	Assessment of findings and clinical significance	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
Report (29501.2)	File name of uploaded copy of clinical report - requested format [Participant ID]_[Local Report Identifier]	0..1	xs:base64Binary

together with, in the case of in the case of

3.3.9.1 Exercise test (30214.1)

Name	Description	Multiplicity	Data Type
long exercise test result (30217.1)	Result of long exercise test	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

Short exercise test result (30218.1)	Result of short exercise test	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition
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or, in the case of

3.3.9.2 Childhood Health Assessment (30220.1)

Name	Description	Multiplicity	Data Type
Childhood Health Assessment score (30219.1)	Result of Childhood Health Assessment using questionnaire	1..1	xs:decimal

or, in the case of

3.3.9.3 Childhood Myositis Assessment (30222.1)

Name	Description	Multiplicity	Data Type
Childhood Myositis Assessment score (30221.1)	Result of Childhood Myositis Assessment	1..1	xs:decimal

or, in the case of

3.3.9.4 Intelligence Quotient Assessment (30224.1)

Name	Description	Multiplicity	Data Type
Intelligence Quotient Score (30223.1)	Result of Intelligence Quotient test	1..1	xs:integer
Intelligence Quotient assessment method (31287.1)	Method used to assess Intelligence Quotient	0..1	xs:string

or, in the case of

3.3.9.5 Performance Intelligence Quotient assessment (30226.1)

Name	Description	Multiplicity	Data Type
Performance intelligence quotient score (30225.1)	Result of Performance intelligence quotient test	1..1	xs:integer
Performance Intelligence Quotient assessment method (31288.1)	Method used for assessment of performance intelligence quotient	0..1	xs:string

or, in the case of

3.3.9.6 Ataxia assessment (30228.2)

Name	Description	Multiplicity	Data Type
Inventory of Non-Ataxia Signs score (30227.2)	Result of INAS ataxia test	1..1	xs:integer
Scale for the Assessment and Rating of Ataxia (SARA) (28907.1)	Scale for the Assessment and Rating of Ataxia (SARA) score	0..1	xs:nonNegativeInteger

or, in the case of

3.3.9.7 Spastic Paraplegia Assessment (30230.1)

Name	Description	Multiplicity	Data Type
Spastic Paraplegia score (30229.1)	Results of Spastic Paraplegia test	1..1	xs:integer
Spastic Paraplegia assessment method (31290.1)	Method used for the assessment of spastic paraplegia	0..1	xs:string

or, in the case of

3.3.9.8 Manual Muscle Testing 8 (MMT8) Assessment (30232.1)

Name	Description	Multiplicity	Data Type
Manual Muscle Testing 8 (MMT8) score (30231.1)	Result of Manual Muscle Testing 8 (MMT8) test	1..1	xs:integer

or, in the case of

3.3.9.9 Visual Analog Pain Scale Assessment (30239.1)

Name	Description	Multiplicity	Data Type
Physician global activity assessment using visual analog pain scale (30238.1)	Global activity pain assessment by physician	0..1	visualAnalogPainScore
Physician extra-muscular global activity assessment using visual analog pain scale (30237.1)	Extra-muscular global activity pain assessment by physician	0..1	visualAnalogPainScore
Patient assessment using visual analog pain scale (30235.1)	Pain assessment by patient	0..1	visualAnalogPainScore
Parental assessment using visual analog pain scale (30234.1)	Pain assessment by parent	0..1	visualAnalogPainScore

or, in the case of

3.3.9.10 Development Quotient Assessment (30244.2)

Name	Description	Multiplicity	Data Type
Development Quotient assessment method (30242.2)	Method used for development quotient assessment	1..1	developmentQuotientMethod <i>Enumeration</i> Griffiths: Griffiths WISC-3: WISC-3 WPPSI: WPPSI Bayley: Bayley
Development Quotient assessment score (30243.1)	Result of development quotient assessment using specified method	1..1	xs:double

or, in the case of

3.3.9.11 Beighton test (31476.1)

Name	Description	Multiplicity	Data Type
Beighton score (28583.1)	A measure assessing hyperflexibility	0..1	xs:decimal

3.3.10 Vital signs (30111.2)

All reports for vital signs should include the following elements,

Name	Description	Multiplicity	Data Type
Assessment (29524.1)	Assessment of findings and clinical significance	0..1	Clinical Test Abnormality <i>Enumeration</i> Normal: Normal Unknown: No results available Abnormal-Relevant: An abnormality of clinical relevance to the patient's condition Abnormal-Unknown Relevance: An abnormality of unknown clinical relevance to the patient's condition

together with, in the case of

3.3.10.1 Blood pressure (30245.1)

Name	Description	Multiplicity	Data Type
Diastolic arterial blood pressure - NBPDiastolic arterial blood pressure (28059.1)	Diastolic (non-invasive) arterial blood pressure measurement	0..1	xs:integer
Systolic Arterial blood pressure - NBPSystolic Arterial blood pressure (28054.1)	Systolic (non-invasive) arterial blood pressure measurement	0..1	xs:integer

or, in the case of

3.3.10.2 Additional body measurements (30247.2)

Name	Description	Multiplicity	Data Type
Father's height (28633.1)	Father's height in cm	0..1	lengthInCm
Mother's height (28632.1)	Mother's height in cm	0..1	lengthInCm

Upper segment height (30254.1)	Height of upper body segment in cm	0..1	lengthInCm
Final Height (30249.1)	Final height of person in cm	0..1	lengthInCm
Height on Presentation (30250.1)	Person's height on presentation in cm	0..1	lengthInCm
Lower segment height (30253.1)	Height of lower body segment in cm	0..1	lengthInCm
Arm span (AS) (28588.1)	Arm span in cm	0..1	lengthInCm
Maternal head circumference (28863.1)	Circumference of mother's head in cm	0..1	headCircumference
Paternal head circumference (28864.1)	Circumference of father's head in cm	0..1	headCircumference
Weight at presentation (30255.1)	Person's weight in kg at presentation	0..1	personObservation(weight)

3.4 Diagnoses (31151.3)

See *diagnosis* (31151.3)

3.5 Intervention (29528.2)

All reports for interventions should include the following elements,

Name	Description	Multiplicity	Data Type
Event Date (12727.4)	Date of the clinical event or observation being reported e.g. date biopsy was taken	1..1	xs:dateTime
Event Reference (14858.3)	Unique identifier for local record of clinical event or observation	0..1	xs:string

together with, in the case of

3.5.1 Medication (29852.1)

Name	Description	Multiplicity	Data Type
Start Date (14961.1)	Start date for the proposed treatment. This may or may not be known at the time of care planning, and therefore is optional.	0..1	xs:date
End Date (12803.1)	End date, if applicable	0..1	xs:date
Drug Code (12799.1)	Dm+d / Snomed CT Code for the medication used. For more information please visit the dm+d browser: http://www.ppa.org.uk/systems/pcddbrowserv2_3new/browser.jsp#product	1..1	snomedCtDm+d
Drug Name (29855.1)	dm+d drug name - without manufacturer	0..1	xs:string
Dose (12800.1)	Dose prescribed according to the dictionary of medications and devices (http://www.nhsbsa.nhs.uk/1121.aspx)	0..1	xs:string
Frequency (12801.1)	Frequency prescribed	0..1	xs:string

or, in the case of

3.5.2 Surgical Procedure (29856.1)

Name	Description	Multiplicity	Data Type
Primary Procedure (SnomedCT) (29858.1)	Snomed CT code for the primary procedure	0..1	procedureCodeSnomedCT
Primary Procedure	OPCS-4 code for the primary	0..1	OPCS-4

(OPCS-4) (30860.1)	procedure		
Additional Procedures (SnomedCT) (12807.1)	If additional procedures, SnomedCT procedure code for procedure	0..1	procedureCodeSnomedCT
Additional Procedures (OPCS-4) (30861.1)	If additional procedures, OPCS-4 procedure code for procedure	0..1	OPCS-4
Comments (14954.1)	Follow-up comments	0..1	xs:string

3.5.3 Other Procedure (29859.1)

Other procedures i.e. those other than surgical procedures or medication.

Name	Description	Multiplicity	Data Type
Start Date (14961.1)	Start date for the proposed treatment. This may or may not be known at the time of care planning, and therefore is optional.	0..1	xs:date
End Date (12803.1)	End date, if applicable	0..1	xs:date
Primary Treatment (OPCS-4) (31145.1)	OPCS-4 code for non-surgical and non-medicinal primary treatment	0..1	OPCS-4
Additional Treatment (OPCS-4) (31146.1)	OPCS-4 code for non-surgical and non-medicinal additional treatments	0..1	OPCS-4
Primary Treatment (SnomedCT) (12812.1)	Snomed CT code for non-surgical and non-medicinal primary treatment	0..1	procedure(snomedCt)
Additional Treatment (SnomedCT) (12813.1)	Snomed CT code for non-surgical and non-medicinal additional treatments	0..unbounded	procedure(snomedCt)

3.6 Withdrawal

Name	Description	Multiplicity	Data Type
Date of withdrawal of consent (14667.2)	The date withdrawal occurred	1..1	xs:date
Name and Version of the Withdrawal Form Used (12729.2)	Name and Version of form used - list of names and versions available from genomicsengland.co.uk/library-and-resources/	1..1	genomicsEnglandConsentWithdrawalForms
Withdrawal Option (12728.1)	Indicating full or partial withdrawal	1..1	consentWithdrawalOptions <i>Enumeration</i> FULL_WITHDRAWAL :OPTION 2: FULL WITHDRAWAL: No further use PARTIAL_WITHDRAWAL :OPTION 1: PARTIAL WITHDRAWAL: No further contact
Withdrawal Form (12730.3)	Filename of uploaded copy of scanned withdrawal form pdf - requested format is [ParticipantId]_withdrawal_[TimeStamp].pdf	1..1	xs:string
Person Reporting Withdrawal (12731.2)	Full name, including forenames and surname, of person reporting withdrawal.	1..1	xs:string

3.7 Death Details (33514.1)

A report of death should include (see NHS data dictionary):

http://www.datadictionary.nhs.uk/data_dictionary/classes/p/person_death_details_at.asp?shownav=1

Name	Description	Multiplicity	Data Type
Event Date (12727.4)	Date of the clinical event or observation being reported e.g. date biopsy was taken	1..1	xs:dateTime
Event Reference (14858.3)	Unique identifier for local record of clinical event or observation	0..1	xs:string
Death Location (12777.1)	Location of death	0..1	deathLocation <i>Enumeration</i> 3: Voluntary hospice / Specialist Palliative Care unit 2: NHS hospice / Specialist Palliative Care unit 1: Hospital 6: Other 5: Care Home 4: PATIENT's own home
Immediate Cause (12778.1)	Immediate cause of death. Coded according to the International Classification of Diseases (ICD) code of the condition leading to death as recorded on the death certificate.	0..1	deathCauseCode
Condition (12780.1)	Condition leading to death. Coded according to the International Classification of Diseases (ICD) code of the condition leading to death as recorded on the death certificate.	0..1	deathCauseCode
Underlying Cause (12779.1)	Underlying cause of death. Coded according to the International Classification of Diseases (ICD) code of the condition leading to death as recorded on the death certificate.	0..1	deathCauseCode
Significant (12781.1)	Significant condition not leading to death. Coded according to the	0..1	deathCauseCode

	International Classification of Diseases (ICD) code of the condition leading to death as recorded on the death certificate.		
Diagnosis (ICD) (33515.1)		0..unbounded	

4 Additional Data (12786.1)

All of the additional data items provided following registration correspond to items in a specific longitudinal record model, in which each piece of data reported is associated with a date, usually the date of a relevant clinical event.

The additional data required for a participant includes data on relevant investigations, diagnoses, and interventions in the existing medical history. A list of relevant events will be provided for each rare disorder.

It includes also additional phenotyping statements: the list of recommended statements for a given disorder will be extended periodically, in response to new insights and discoveries, leading to additional questions for existing participants.

Coded values will be accepted against ODS, OPCS, ICD, and Snomed-CT standards. Existing data should be reported in the form in which it was originally recorded; GMCs are encouraged to use Snomed-CT for new data where possible.

The additional data for a rare disease participant should be supplied within four weeks of the event in question.

All additional data will be recorded against the same data model as that set out for core data in Section 3 above.

Additional Data model does not have any child models or data elements yet.

5 Value Domains

Axis

(Genomics England Data Set)

Data Type	xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.
Unit of Measure	Degrees (Measure of an angle)

Body Region (face)

(Genomics England Data Set)

Data Type	Body Region (face)
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Enumeration

Code	Description
Head	Head

Body Region (heart)

(Genomics England Data Set)

Data Type	Body Region (heart)
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Enumeration

Code	Description
Heart	Heart

Body Region (heart/liver)

(Genomics England Data Set)

Data Type Body Region (heart/iron)

Enumeration

Code	Description
Heart	Heart
Liver	Liver

Body System (heart)

(Genomics England Data Set)

Data Type Body System (heart)

Enumeration

Code	Description
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Cardiovascular	Cardiovascular
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Body System (heart/liver)

(Genomics England Data Set)

Data Type Body System (heart/liver)

Enumeration

Code	Description
Cardiovascular	Cardiovascular
Digestive	Digestive

C3 complement

(Genomics England Data Set)

Data Type highLowNormal

Enumeration

Code	Description
High	Raised above normal level
Low	Reduced below normal level

Normal	At a normal level
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C4 complement

(Genomics England Data Set)

Data Type highLowNormal

Enumeration

Code	Description
High	Raised above normal level
Low	Reduced below normal level
Normal	At a normal level

Clinical Test Abnormality

(Genomics England Data Set)

Clinical Test Abnormality

Data Type Clinical Test Abnormality

Enumeration

Code	Description
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Normal	Normal
Unknown	No results available
Abnormal-Relevant	An abnormality of clinical relevance to the patient's condition
Abnormal-Unknown Relevance	An abnormality of unknown clinical relevance to the patient's condition

Conception

(Genomics England Forms)

Data Type

Conception

Enumeration

Code	Description
Spontaneous	Spontaneous
Other	Other
InVitro	In vitro fertilisation

Consanguinity

(Genomics England Data Set)

This is an indicator of whether a person is the product of a consanguinous relationship

Data Type YesNoUnkPos (Yes, No, Possible, Unknown)

Enumeration

Code	Description
U	Unknown
P	Possible
N	No
Y	Yes

FVCpercentage

(Genomics England Data Set)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Percentage

HFLA

(Genomics England Data Set)

HFLA for signal averaged ECG

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure microVolts (Measurement of potential equal to one millionth of a volt)

ICD10Code

(ICD)

Data Type icdCode

Imaging Modality (Doppler Diastolic Function)

(Genomics England Data Set)

Data Type Imaging Modality (Doppler Diastolic function)

Enumeration

Code	Description
Doppler Diastolic Function Assessment	Doppler Diastolic Function Assessment

Imaging Modality (Echocardiogram)

(Rare Diseases)

Data Type Imaging Modality (Echocardiogram)

Enumeration

Code	Description
Diagnostic Ultrasonography	Diagnostic Ultrasonography
Other	Other

Imaging Modality (Kidney Imaging)

(Genomics England Data Set)

Data Type Imaging Modality (Kidney Imaging)

Enumeration

Code	Description
Computerised Axial Tomography	Computerised Axial Tomography
Diagnostic Ultrasonography	Diagnostic Ultrasonography
Magnetic Resonance Imaging	Magnetic Resonance Imaging
Nuclear Medicine	Nuclear Medicine
Plain Radiography	Plain Radiography
Positron Emission Tomography	Positron Emission Tomography
Single Photon Emission Computed Tomography	Single Photon Emission Computed Tomography
SAP scan	SAP scan

Other	Other
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Imaging Modality (heart MRI)

(Genomics England Data Set)

Data Type Imaging Modality (heart MRI)

Enumeration

Code	Description
Magnetic Resonance Imaging	Magnetic Resonance Imaging

Imaging Modality (heart/liver)

(Genomics England Data Set)

Data Type Imaging Modality (heart/liver)

Enumeration

Code	Description
Magnetic Resonance Imaging	Magnetic Resonance Imaging
Other	Other

Imaging Procedure (Liver)

(Genomics England Data Set)

Data Type

Imaging Procedure (Liver)

Enumeration

Code	Description
Computerised Axial Tomography	Computerised Axial Tomography
Diagnostic Ultrasonography	Diagnostic Ultrasonography
Magnetic Resonance Imaging	Magnetic Resonance Imaging
Nuclear Medicine	Nuclear Medicine
Plain Radiography	Plain Radiography
Positron Emission Tomography	Positron Emission Tomography
Single Photon Emission Computed Tomography	Single Photon Emission Computed Tomography
SAP scan	SAP scan
Other	Other

Imaging Submodality (Echocardiogram)

(Rare Diseases)

Data Type

Imaging Submodality (Echocardiogram)

Enumeration

Code	Description
Echocardiogram	Echocardiogram
Other	Other

Imaging Submodality (Kidney Imaging)

(Genomics England Data Set)

Data Type Imaging Submodality (Kidney Imaging)

Enumeration

Code	Description
DEXA	dual energy X-ray absorptiometry
DMSA	dimercaptosuccinic acid scan
Micturating cystourethrogram	Micturating cystourethrogram

Imaging Submodality (Liver)

(Genomics England Data Set)

Data Type Imaging Submodality (Liver)

Enumeration

Code	Description
DEXA	dual energy X-ray absorptiometry
DMSA	dimercaptosuccinic acid scan
Ferriscan	Ferriscan
T2*MRI	T2*MRI

Imaging Submodality (heart MRI)

(Genomics England Data Set)

Data Type Imaging Submodality (heart MRI)

Enumeration

Code	Description
Cardiac MRI	Cardiac MRI

Imaging Submodality (heart/liver)

(Genomics England Data Set)

Data Type Imaging Submodality (heart/liver)

Enumeration

Code	Description
Ferriscan	Ferriscan
T2*MRI	T2*MRI

Laterality

(Human Phenotype Ontology)

The localization with respect to the side of the body of the specified phenotypic abnormality.

Data Type Laterality (The localization with respect to the side of the body of the specified phenotypic abnormality.)

Enumeration

Code	Description
Right	Being located on the right side of the body.
Unilateral	Being present on only the left or only the right side of the body.
Bilateral	Being present on both sides of the body.
Left	Being located on the left side of the body.

LowNormalHighNotRecorded

Data Type Low_Normal_High_NotRecorded

Enumeration

Code	Description
Not Recorded	Not Recorded
Low	Low
Normal	Normal
High	High

Months

(SI)

Length of time in months

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Month (Length of time in Months where there are 12 months in a Year

Nasal Cilia Procedure

(Rare Diseases)

Data Type Nasal Cilia Procedure

Enumeration

Code	Description
Biopsy or scrape of nasal cilia	Biopsy or scrape of nasal cilia
Other	Other

OFC

(Genomics England Forms)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure centimeters

OPCS-4

(OPCS)

OPCS-4

Data Type OPCS-4

Onset

(Human Phenotype Ontology)

The age group in which disease manifestations appear. Comment: Adolescent is defined by WHO as a person between 10-19 years of age.

Data Type Onset (The age group in which disease manifestations appear. Comment: Adolescent is defined by WHO as a person between 10-19 years of age.

Enumeration

Code	Description
Embryonal onset	Onset of disease at up to 8 weeks of gestation.
Fetal onset	Onset prior to birth but after 8 weeks of gestation.
Neonatal onset	Onset of signs or symptoms of disease within the first 28 days of life.
Infantile onset	Onset of signs or symptoms of disease between 28 days to one year of life. Comment: Onset of signs or symptoms of disease within the first 24 months of life.
Childhood onset	Onset of disease at the age of between 1 and 5 years.
Juvenile onset	Onset of signs or symptoms of disease between the age of 5 and 15 years.
Young adult onset	Onset of disease at the age of between 16 and 40 years.
Late Onset	A type of adult onset with onset of symptoms after the age of 60 years.
Middle age onset	A type of adult onset with onset of symptoms at the age of 40 to 60 years. Comment: Middle age is of course not amenable to precise definitions. We suggest using this term for onset of disease symptoms between the age of 40 and 60 years.

(Genomics England Data Set)

Data Type PassFail

Enumeration

Code	Description
Fail	Fail
Pass	Pass

PositiveNegativeUnknown

(Genomics England Data Set)

Positive negative or unknown result

Data Type posNegUnk (Positive, negative or unknown

Enumeration

Code	Description
unknown	Unknown
negative	Negative
positive	Positive

Postcode

(PDS)

The UK format Postcode, 8 character string, as per BS7666. The 8 characters field allows a space to be inserted to differentiate between the inward and outward segments of the code, enabling full use to be made of Royal Mail postcode functionality.

N.B. Must be capitalized

Data Type xs:string (Character strings in XML.

Regular ^([GIR ?0AA|[A-PR-UWYZ]([0-9]{1,2}|([A-HK-Y][0-9]([0-9ABEHMNPRV-Y])?)|[0-9][A-HJKPS-UW])
Expression ?[0-9][ABD-HJLNP-UW-Z]{2})\$

Present

(Genomics England Data Set)

Presence of the phenotypic abnormality.

Data Type Phenotype_Present (Presence of the phenotypic abnormality.

Enumeration

Code	Description
unknown	Unknown
yes	Yes
no	No

PresentAbsent

(Genomics England Data Set)

Data Type PresentAbsent

Enumeration

Code	Description
Present	Present
Absent	Absent

Progression

(Genomics England Data Set)

Data Type Progression

Enumeration

Code	Description
Progressive	Progressive
Nonprogressive	Nonprogressive

RMS

(Genomics England Data Set)

RMS value from signal average ECG

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure microVolts (Measurement of potential equal to one millionth of a volt

Region (Echocardiogram)

(Rare Diseases)

Region (Echocardiogram)

Data Type Region (Echocardiogram) (Region (Echocardiogram)

Enumeration

Code	Description
Heart	Heart

Renal Biopsy Procedure

(Rare Diseases)

Data Type Renal Biopsy Procedure

Enumeration

Code	Description
Kidney biopsy	Kidney biopsy
Other	Other

Severity

(Human Phenotype Ontology)

The intensity or degree of a manifestation.

Data Type

Severity (The intensity or degree of a manifestation.)

Enumeration

Code	Description
Borderline	Having a minor degree of severity that is considered to be on the boundary between the normal and the abnormal ranges. For quantitative traits, a deviation of that is less than two standard deviations from the appropriate population mean.
Mild	Having a relatively minor degree of severity. For quantitative traits, a deviation of between two and three standard deviations from the appropriate population mean.
Moderate	Having a medium degree of severity. For quantitative traits, a deviation of between three and four standard deviations from the appropriate population mean.
Severe	Having a high degree of severity. For quantitative traits, a deviation of between four and five standard

	deviations from the appropriate population mean.
Profound	Having an extremely high degree of severity. For quantitative traits, a deviation of more than five standard deviations from the appropriate population mean.

System (Echocardiogram)

(Rare Diseases)

System (Echocardiogram)

Data Type System (Echocardiogram) (System (Echocardiogram))

Enumeration

Code	Description
Cardiovascular	Cardiovascular

Weeks

(SI)

Length of time in weeks

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Week (Measurement of time where there are seven days in a week.

adaptation

(Genomics England Data Set)

Data Type darkLight

Enumeration

Code	Description
dark	Dark adaptation used in ERG
light	Light adaptation used in ERG

addressLine

(PDS)

Includes main, temporary and correspondence addresses
5 lines excludes postcode, may be vernacular or PAF derived. The following address lines should normally be present although there may be some exceptions:
-1 or 2,
-and 4

Data Type xs:string (Character strings in XML.

Rule maxLength(175)

adoptedStatus

(Annex D)

Data Type adoptedStatus

Enumeration

Code	Description
not_adopted	not adopted
adopted_in	adopted into the family
adopted_out	child belonged to the family and was adopted out

affectedStatus

(Genomics England Data Set)

Is this person affected

Data Type yesNoUnk

Enumeration

Code	Description
yes	Yes
no	No
unknown	Unknown

ageOfOnsetYears

(Genomics England Data Set)

Age of onset measured in years

Data Type xs:nonNegativeInteger (Infinite set {0, 1, 2,...}. Sign omitted, “+” assumed. Example: 1, 0, 12678967543233, +100000.

Unit of Measure Year

amplitude

(Genomics England Data Set)

amplitude measurement in microvolts, developed for ocular measurement readings

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure microVolts (Measurement of potential equal to one millionth of a volt

analysedSpecimenType

(Rare Diseases)

Data Type analysedSpecimenType

Enumeration

Code	Description
------	-------------

Air	Air sample
Amniotic fluid	Amniotic fluid specimen
Ascitic fluid	Ascitic fluid sample
BAL	Bronchoalveolar lavage fluid sample
Bile	Bile specimen
Blood	Blood specimen
Blood spot	Blood spot specimen
Body fluid	Body fluid sample
Bone marrow	Bone marrow specimen
Breast milk	Expressed breast milk specimen
Breath	Breath sample
Calculus	Calculus specimen
Cells	Cell sample
CSF	Cerebrospinal fluid sample
Dialysis fluid	Dialysis fluid specimen
ETA	Endotracheal aspirate (specimen)
Faeces	Stool specimen

Fibroblasts	Fibroblast specimen
Food item	Food specimen
Foreign body	Foreign body submitted as specimen
Hair	Hair specimen
Medical device	Device specimen
Nail	Nail specimen
NPA	Nasopharyngeal aspirate
Pericardial fluid	Pericardial fluid specimen
Plasma citrate	Plasma specimen with citrate
Plasma EDTA	Plasma specimen with EDTA (ethylenediaminetetraacetic acid)
Plasma fluoride oxalate	Plasma specimen with fluoride oxalate
Plasma lithium heparin	Plasma specimen with lithium heparin
Platelets	Platelet specimen
Pleural fluid	Pleural fluid specimen
Prosthetic material	Specimen from prosthetic device
Pus	Pus specimen
Red cells	Erythrocyte specimen

Saliva	Saliva specimen
Semen	Semen specimen
Serum	Serum specimen
Skin	Skin (tissue) specimen
Sputum	Sputum specimen
Swab	Swab
Sweat	Sweat specimen
Synovial fluid	Synovial fluid specimen
Tip	Tip - central venous ***
Catheter	Catheter (CVC)
Tip - Other	Tip - Other Device tip sample
Tissue	Tissue specimen
Urine	Urine specimen
Vitreous fluid	Vitreous humour sample
Vomit	Vomit specimen
Water	Water specimen
White cells	White blood cell sample

Whole blood EDTA	Whole blood specimen with EDTA (ethylenediaminetetraacetic acid)
Whole blood fluoride oxalate	Whole blood specimen with fluoride oxalate
Whole blood heparinised	Whole blood specimen with heparin
Whole blood perchlorate	Whole blood specimen with perchlorate
Other	Other

analysedSpecimenType Arterialbloodgas

Data Type analysedSpecimenType Arterialbloodgas

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Autoantibodies

Data Type analysedSpecimenType Autoantibodies

Enumeration

Code	Description
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Blood	Blood
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analysedSpecimenType BloodTests

Data Type analysedSpecimenType BloodTests

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Boneprofile

Data Type analysedSpecimenType Boneprofile

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType CSFtests

Data Type analysedSpecimenType CSFtests

Enumeration

Code	Description
CSF	CSF

analysedSpecimenType Fecaltest

Data Type analysedSpecimenType Fecaltest

Enumeration

Code	Description
Faeces	Faeces

analysedSpecimenType FullBloodCount

Data Type analysedSpecimenType FullBloodCount

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Liverbiochemistry

Data Type analysedSpecimenType Liverbiochemistry

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Otherenzymes

Data Type analysedSpecimenType Otherenzymes

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Pancreaticautoantibodies

Data Type analysedSpecimenType Pancreaticautoantibodies

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Renalbiochemistry

Data Type analysedSpecimenType Renalbiochemistry

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType Serumimmunoglobulins

Data Type analysedSpecimenType Serumimmunoglobulins

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType TORCHscreen

Data Type analysedSpecimenType TORCHscreen

Enumeration

Code	Description
Blood	Blood

analysedSpecimenType UrineDip

Data Type analysedSpecimenType UrineDip

Enumeration

Code	Description
Urine	Urine

analysedSpecimenType UrineTest

Data Type analysedSpecimenType UrineTest

Enumeration

Code	Description
Urine	Urine

antibodyName

(Genomics England Data Set)

Data Type antibodyNames

Enumeration

Code	Description
ANA	ANA
ANCA	ANCA
Anti-p155/140	Anti-p155/140
Anti-p140	Anti-p140
Anti-Jo1	Anti-Jo1
Anti-ARS	Anti-ARS
Anti-PL12	Anti-PL12
Anti-PL7	Anti-PL7
Anti-EJ	Anti-EJ
Anti-OJ	Anti-OJ
Anti-Mi2	Anti-Mi2
Anti-SRP	Anti-SRP
Anti-U1-RNP	Anti-U1-RNP
Anti-U3-RNP	Anti-U3-RNP
Anti-PM-Scl 75	Anti-PM-Scl 75
Anti-PM-Scl 100	Anti-PM-Scl 100

Anti-Ro52	Anti-Ro52
Anti-Ro60	Anti-Ro60
Anti-La	Anti-La
Anti-Ku	Anti-Ku
Anti-Topo	Anti-Topo
Anti-SAE	Anti-SAE
Anti-Zo	Anti-Zo
Anti-CAM-140	Anti-CAM-140
Anti-KS	Anti-KS
Anti-HA	Anti-HA
Anti-ZA	Anti-ZA
Other MSA / MAA	Other MSA / MAA
Anti-dsDNA	Anti-dsDNA
Cryoglobulins	Cryoglobulins
cANCA	cANCA
ENA	ENA
Hepatitis B Antibody	Hepatitis B Antibody

Hepatitis C Antigen	Hepatitis C Antigen
HIV Antibody	HIV Antibody
pANCA	pANCA
Other	Other

antibodyResult

(Genomics England Data Set)

Data Type posNegUnkNot

Enumeration

Code	Description
positive	Positive
negative	Negative
unknown	Unknown significance
not done	Not done

apgarScore

(Genomics England Data Set)

Assessment of health of a newborn

Data Type xs:nonNegativeInteger (Infinite set {0, 1, 2,...}. Sign omitted, “+” assumed. Example: 1, 0, 12678967543233, +100000.

areaInCm2

(Genomics England Data Set)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Square centimetre (Unit of area

auditoryBrainstemResponseResult

(Genomics England Data Set)

Data Type auditoryBrainstemResponseResult (Result of ABR test

Enumeration

Code	Description
Normal	Normal
Abnormal	Abnormal
Abnormal with no evidence of peaks	Abnormal with no evidence of peaks
Abnormal with early evidence of peaks (up to wave III)	Abnormal with early evidence of peaks (up to wave III)
Abnormal morphology but wave V present	Abnormal morphology but wave V present

basementMembrane

(Genomics England Data Set)

Describes the result of an examination of renal basement membrane

Data Type normalThinnedAbnormal

Enumeration

Code	Description
normal	Normal
thinned	Thinned
abnormal	Abnormal

beatFrequencyInHz

(Genomics England Data Set)

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure Hertz

biologicalFamilyRelationship

(Genomics England Data Set)

Biological Family Relationship

Data Type BiologicalFamilyRelationship (Biological Family Relationship between two people)

Enumeration

Code	Description
Mother	Mother
Father	Father
TwinsMonozygous	Twins-Monozygous
TwinsDizygous	Twins-Dizygous
TwinsUnknown	Twins-Unknown
FullSibling	FullSibling
FullSiblingM	Half Sibling (shared mother)
FullSiblingF	Half Sibling (shared father)
MaternalAunt	Maternal Aunt
PaternalAunt	Paternal Aunt
MaternalUncle	Maternal Uncle
PaternalUncle	Paternal Uncle
MaternalCousinSister	Maternal Cousin - child of mother's sister

MaternalCousinBrother	Maternal Cousin - child of mother's brother
PaternalCousinSister	Paternal Cousin - child of father's sister
PaternalCousinBrother	Paternal Cousin - child of father's brother
MaternalGrandmother	Maternal Grandmother
MaternalGrandfather	Maternal Grandfather
PaternalGrandmother	Paternal Grandmother
PaternalGrandfather	Paternal Grandfather
DoubleFirstCousin	Double First Cousin
FirstCousinOnceRemoved	First Cousin OnceRemoved
SecondCousin	Second Cousin
SecondCousinOnceRemoved	Second Cousin Once Removed
ThirdCousin	Third Cousin
Son	Son
Daughter	Daughter
Other	Other (Please specify)

biopsyAnalysisType

(Genomics England Data Set)

Biopsy Analysis Type

Data Type biopsyAnalysisType (Type of biopsy analysis)

Enumeration

Code	Description
Light Microscopy	Light Microscopy
Immunohistochemistry	Immunohistochemistry
Electron Microscopy	Electron Microscopy
Biochemistry	Biochemistry
Respiratory chain enzyme analysis	Respiratory chain enzyme analysis

biopsyProcedure

(Genomics England Data Set)

Data Type biopsyProcedure

Enumeration

Code	Description
Biopsy of peripheral nerve	Biopsy of peripheral nerve

Biopsy of peritoneum	Biopsy of peritoneum
Biopsy of skin and/or subcutaneous tissue	Biopsy of skin and/or subcutaneous tissue
Biopsy of skin for fibroblast culture	Biopsy of skin for fibroblast culture
Biopsy or scrape of nasal cilia	Biopsy or scrape of nasal cilia
Biopsy of skeletal muscle	Biopsy of skeletal muscle
Bone marrow biopsy, needle or trocar	Bone marrow biopsy, needle or trocar
Kidney biopsy	Kidney biopsy
Transplant kidney biopsy	Transplant kidney biopsy
Other	Other

bodyRegion

(Rare Diseases)

Data Type bodyRegion (Based on Region categories derived from submitted NICIP or SNOMED-CT codes. The following category descriptions have been defined:

Enumeration

Code	Description
Abdominal	Abdominal
Head	Head

Heart	Heart
Inner ear	Inner ear
Limb	Limb
Liver	Liver
Neck	Neck
Outer ear	Outer ear
Pelvic	Pelvic
Spine	Spine
Thoracic	Thoracic
Whole body	Whole body
None	None
Unknown	Unknown

bodyStructure

(Genomics England Data Set)

snomedCt subclassifications of body system

Data Type

snomedCt

bodySystem

(Genomics England Data Set)

The body system investigated by a test

Data Type bodySystem (Body systems

Enumeration

Code	Description
Auditory	Auditory
Cardiovascular	Cardiovascular
Digestive	Digestive
Endocrine	Endocrine
Genitourinary	Genitourinary
Haematological	Haematological
Lymphatic	Lymphatic
Lymphoreticular	Lymphoreticular
Musculoskeletal	Musculoskeletal
Nervous	Nervous
Respiratory	Respiratory
Visual	Visual

Unknown	Unknown
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calculatedDrugAmount

(Genomics England Data Set)

Calculated drug amount for cardiac drug challenge test

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure milligrams (A unit of mass equal to one thousandth (10⁻³) of a gram

cellCountUnitOfMeasurement

(Genomics England Data Set)

Data Type cellCount

Enumeration

Code	Description
number times ten raised to the power of nine per litre (x10 ⁹ /l)	number times ten raised to the power of nine per litre (x10 ⁹ /l)

chiNumber

(NHS Data Dictionary)

The Community Health Index (CHI) is a population register, which is used in Scotland for health care purposes. The CHI number uniquely identifies a person on the index.

Data Type	xs:string (Character strings in XML.
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Rule	length(10)
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concentrationInPmol/l

(Genomics England Data Set)

Data Type	xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes optional. If the fractional part is zero, the period and following zero(es) can be omitted.
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Unit of Measure	picomole/litre
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concentrationMmol/l

(Genomics England Data Set)

Data Type	xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes optional. If the fractional part is zero, the period and following zero(es) can be omitted.
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Unit of Measure	concentration mmol/l (substance concentration in mmols per litre
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concentrationUnitOfMeasurement

(Genomics England Data Set)

Data Type	concentration
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Enumeration

Code	Description
Grams per decilitre (g/dl)	Grams per decilitre (g/dl)
Grams per litre (g/l)	Grams per litre (g/l)
International Units per litre (IU/L)	International Units per litre (IU/L)
Micrograms per litre (µg/L)	Micrograms per litre (µg/L)
Micrograms per millilitre (µg/ml)	Micrograms per millilitre (µg/ml)
Micrograms per millimole (µg/mmol)	Micrograms per millimole (µg/mmol)
Micromoles per litre (µmol/L)	Micromoles per litre (µmol/L)
Milligrams per litre (mg/l)	Milligrams per litre (mg/l)
Millimoles per litre (mmol/L)	Millimoles per litre (mmol/L)
Nanograms per millilitre (ng/ml)	Nanograms per millilitre (ng/ml)
Picomoles per litre (pmol/L)	Picomoles per litre (pmol/L)

consentWithdrawalOptions

(Genomics England Data Set)

Genomics England Consent Withdrawal Options

Data Type

ConsentWithdrawalOptions (Genomics England Consent Withdrawal Options)

Enumeration

Code	Description
FULL_WITHDRAWAL	OPTION 2: FULL WITHDRAWAL: No further use
PARTIAL_WITHDRAWAL	OPTION 1: PARTIAL WITHDRAWAL: No further contact

cornealCurvature

(Genomics England Data Set)

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure Dioptre

deathCauseCode

(Genomics England Data Set)

DEATH CAUSE ICD CODE is the International Classification of Diseases (ICD) code of the condition leading to death as recorded on the death certificate.

Data Type xs:string (Character strings in XML.

deathLocation

(Rare Diseases)

The type of LOCATION at which a PERSON died.

For the purposes of the Community Information Data Set this is either the LOCATION where the PATIENT expressed a preference to die, or where they actually died.

deathLocationType
([Http://www.datadictionary.nhs.uk/data_dictionary/attributes/d/death_location_type_de.asp?shownav=1](http://www.datadictionary.nhs.uk/data_dictionary/attributes/d/death_location_type_de.asp?shownav=1))

Data Type The type of LOCATION at which a PERSON died.

For the purposes of the Community Information Data Set this is either the LOCATION where the PATIENT expressed a preference to die, or where they actually died.

Enumeration

Code	Description
3	Voluntary hospice / Specialist Palliative Care unit
2	NHS hospice / Specialist Palliative Care unit
1	Hospital
6	Other
5	Care Home
4	PATIENT's own home

degreesAngle

(Genomics England Data Set)

Measurement in degrees of an angle

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Degrees (Measure of an angle

developmentQuotientMethod

(Genomics England Data Set)

Data Type developmentQuotientMethod

Enumeration

Code	Description
Griffiths	Griffiths
WISC-3	WISC-3
WPPSI	WPPSI
Bayley	Bayley

diagnosticTermsSnomedCT

(SNOMED CT)

Data Type snomedCt

diameterInMm

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Millimetres

dopplerDiastolic

(Genomics England Data Set)

Doppler Diastolic function assessment result

Data Type dopplerDiastolic (Doppler Diastolic function assessment results

Enumeration

Code	Description
Normal	Normal
Type 1	Type 1
Type 2	Type 2
Type 3	Type 3
Type 4	Type 4

drugDose

(Genomics England Data Set)

The quantity of a drug administered

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure milligrams (A unit of mass equal to one thousandth (10⁻³) of a gram

durationOfInflammation

(Genomics England Data Set)

Duration of inflammation attacks

Data Type xs:integer (Integer or whole numbers - Sign omitted, “+” is assumed. Example: -1, 0, 12678967543233, +12678967543233)

Unit of Measure days (time in days

electronDenseDeposits

(Genomics England Data Set)

Data Type electronDenseDeposits

Enumeration

Code	Description
mesangial	Mesangial
subendothelial	Subendothelial
subepithelial	Subepithelial

intramembranous	Intramembranous
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electroretinogramResponse

(Genomics England Data Set)

Data Type reducedExtinguished (result indicating reduced or extinguished response)

Enumeration

Code	Description
reduced	Reduced
extinguished	Extinguished

emailAddress

(Genomics England Data Set)

A Valid Email Address i.e. someone@somedomain.com

Data Type xs:string (Character strings in XML.

Regular Expression ^[\w-\.\.]+\@([\w-]+\.\.)+[\w-]{2,4}\$

epinephrineProtocol

(Genomics England Data Set)

Epinephrine cardiac drug challenge protocol types

Data Type epinephrineProtocol (Epinephrine cardiac drug challenge protocol types)

Enumeration

Code	Description
Shimizu	Shimizu
Mayo clinic	Mayo clinic
Other	Other

ethnicCategory

(Cancer Outcomes and Services Dataset)

The ethnicity of a PERSON, as specified by the PERSON.. The 16+1 ethnic data categories defined in the 2001 census is the national mandatory standard for the collection and analysis of ethnicity. (The Office for National Statistics has developed a further breakdown of the group from that given, which may be used locally.)

Data Type EthnicCategory (The ethnicity of a PERSON, as specified by the PERSON.. The 16+1 ethnic data categories defined in the 2001 census is the national mandatory standard for the collection and analysis of ethnicity.
(The Office for National Statistics has developed a further breakdown of the group from that given, which may be used locally.)

Rule maxLength(2)

Enumeration

Code	Description
D	Mixed: White and Black Caribbean
E	Mixed: White and Black African
F	Mixed: White and Asian
G	Mixed: Any other mixed background
A	White: British
B	White: Irish
C	White: Any other White background
L	Asian or Asian British: Any other Asian background
M	Black or Black British: Caribbean
N	Black or Black British: African
H	Asian or Asian British: Indian
J	Asian or Asian British: Pakistani
K	Asian or Asian British: Bangladeshi
P	Black or Black British: Any other Black background
S	Other Ethnic Groups: Any other ethnic group
R	Other Ethnic Groups: Chinese

Z	Not stated
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exerciseStatus

(Genomics England Data Set)

The exercise status of a patient

Data Type ExerciseLevel

Enumeration

Code	Description
Low	Low
Moderate	Moderate
Intense	Intense

fQRSd

(Genomics England Data Set)

fQRSd for signal averaged ECG

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Millisecond

familyAffectionPattern

(Genomics England Data Set)

Pattern of affected individuals in the family

Data Type familyAffectionPattern

Enumeration

Code	Description
singleUnknown	Single affected person with unknown family history
singleIsolated	Single isolated case of disease with unaffected parents
multipleSibsNotParents	Multiple affected full siblings with unaffected parents
multipleNotAllSibs	Multiple affected related individuals, not all of whom are full siblings

fetalBodyStructuresSnomedCt

(Rare Diseases)

fetal body structures snomedCT

Data Type snomedCt

gelSampleProtocolVersion

(Annex D)

Version of the Genomics England Protocol used for sample handling and processing

Data Type gelSampleProtocolVersion (Version of the Genomics England Protocol used for sample handling and processing)

Enumeration

Code	Description
v1	v1 of the Genomics England sample handling and processing protocol
v2	v2 of the Genomics England sample handling and processing protocol

geneScope

(Rare Diseases)

The gene or genes considered

Data Type hgncSymbol (HGNC (HUGO Gene Nomenclature Committee) approved gene name and symbol (short-form abbreviation))

 <http://www.genenames.org/>

geneticTestLaboratory

(Genomics England Data Set)

Was genetic testing performed in a diagnostic or research laboratory?

Data Type genetic_test_laboratory (Was genetic testing performed in a diagnostic or research laboratory?)

Enumeration

Code	Description
research_laboratory	Research laboratory
diagnostic_laboratory	Diagnostic laboratory

geneticTestMethod

(Annex C)

Data Type geneticTestMethod (Genetic Test Method)

Enumeration

Code	Description
Sequencing	Sequencing
Sequencing and targeted copy number analysis	Sequencing and targeted copy number analysis
Copy number analysis	Copy number analysis
Other SNV analysis e.g. OLA	Other SNV analysis e.g. OLA
Targeted copy number analysis e.g. MLPA/qPCR	Targeted copy number analysis e.g. MLPA/qPCR
aCGH	aCGH
SNP array	SNP array

Karyotype	Karyotype
FISH	FISH
Methylation testing	Methylation testing
Microsatellite analysis	Microsatellite analysis
Fanconi (MMC/DEB) breakage testing	Fanconi (MMC/DEB) breakage testing
Radiation hypersensitivity (AT)	Radiation hypersensitivity (AT)
UV hypersensitivity	UV hypersensitivity
Unscheduled DNA synthesis	Unscheduled DNA synthesis
Single Gene Sequencing	Single Gene Sequencing
Gene Panel	Gene Panel
IHC	IHC
Translocation eg qPCR/sequencing/FISH/IHC	Translocation eg qPCR/sequencing/FISH/IHC
Other	Other

geneticTestResult

(Genomics England Data Set)

(for molecular results) If no defect was observed please report 'normal'; if a mutation is detected that is considered pathogenically or clinically important record 'mutation detected'; if no reliable result could be determined please report 'fail'.

Data Type MolecularTestResult (If no defect was observed please report 'normal'; if a mutation is detected that is considered pathogenically or clinically important record 'mutation detected'; if no reliable result could be determined please report 'fail'.

Enumeration

Code	Description
Normal	Normal (negative)
Fail	Fail
AbnormalityDetected	Pathogenic abnormality detected
VUS	Variant of unknown significance detected

genomicsEnglandConsentForms

(Genomics England Data Set)

List of consent forms used by Genomics England

Data Type xs:string (Character strings in XML.

genomicsEnglandConsentWithdrawalForms

(Rare Diseases)

List of consent withdrawal forms used by Genomics England

Data Type xs:string (Character strings in XML.

genomicsEnglandParticipantInformationSheets

(Genomics England Data Set)

Constrained set of names and versions of the participant information sheets that accompany consent

Data Type xs:string (Character strings in XML.

glucoseDipResult

(NHIC TRA)

Data Type xs:string

Unit of Measure mmol/L (mmol/L

gmcFamilyId

A locally-allocated family identifier, unique to this duo or trio.

Data Type xs:string (Character strings in XML.

Regular Expression [0-9]{9}

gmcSampleType

(Rare Diseases)

GMC Sample Type

Data Type

SampleType (The type of the sample (against specified enumeration))

Enumeration

Code	Description
DNA Blood Germline	DNA Blood Germline (CONSTITUTIONAL DNA)
DNA Saliva	DNA Saliva (CONSTITUTIONAL DNA)
DNA Fibroblast	DNA Fibroblast (CONSTITUTIONAL DNA)
DNA FF Germline	DNA FF Germline (CONSTITUTIONAL DNA) - non tumour tissue
DNA FFPE Tumour	DNA FFPE Tumour (TUMOUR DNA)
DNA FF Tumour	DNA FF Tumour (TUMOUR DNA)
DNA Blood Tumour	DNA Blood from blood in Haematological malignancy Tumour (TUMOUR DNA)
EDTA Plasma	EDTA Plasma - Plasma for ctDNA (OMICS SAMPLES)
LiHep Plasma	LiHep Plasma (OMICS SAMPLES)
Tumour Scrapings	FFPE Tumour scrapings or slides (OMICS SAMPLES)
Serum	Serum (OMICS SAMPLES)
RNA Blood	RNA Blood (OMICS SAMPLES)
Buffy Coat	Buffy Coats (OMICS SAMPLES)

Lysed Tumour Cells	Deparaffinised Lysed Tumour Cells in RNA-stabilised buffer (OMICS SAMPLES)
Additional Tumour Material	Additional Tumour material for research (OMICS SAMPLES)

gmcTestResults

(Rare Diseases)

GMC Constrained List of Tests

Data Type GMCLaboratoryTests (GMC Constrained List of Tests)

Enumeration

Code	Description
Agarose	Agarose (Pass / Fail)
Cellularity	The total number of cells (neoplastic and non-neoplastic) in the tumour sample from which DNA was harvested (VeryLow Low Medium High VeryHigh)
delta Cq	delta Cq
Nanodrop OD 260/280	Nanodrop OD 260/280
Nanodrop concentration	Nanodrop concentration ng/ul
Percent Necrosis	Proportion of total number of tumour cells that are neoplastic in the macrodissected tumour and/or sample submitted

Picogreen Concentration	Picogreen Concentration ng/ul
PicoDrop Concentration	PicoDrop Concentration ng/ul
PicoDrop OD 260/280	PicoDrop OD 260/280
Qubit	Qubit ng/ul
Summary QC	Summary QC Pass / Fail
Trinean Concentration	Trinean Concentration ng/ul
Tumour Content	Proportion of total number of tumour cells that are neoplastic (Low, Medium, High) - Low <40%; Medium 40-60%; High >60%.
Trinean OD 260/280	Trinean OD 260/280

groupType

(Genomics England Data Set)

Family members recruited to the study. This will match the categories set out in the 'Guidelines for Family and Proband Selection document'. Codes to follow on release of finalised document.

Data Type TrioDuoSingletonTypes (Constrained list of family study types i.e. whether a duo or trio will be formed)

Enumeration

Code	Description
1	Trio with Mother AND Father

2	Trio with Mother OR Father AND other biological relative
3	Trio with other biological relatives
4	Duo with Mother OR Father
5	Duo with other biological relative
6	Families with more than 3 participants
7	Singleton

hbDipResult

(Genomics England Data Set)

Data Type

hbDipResultType

Enumeration

Code	Description
Negative	Negative
Non-haemolysed trace	Non-haemolysed trace
Haemolysed trace	Haemolysed trace
Small	Small
Moderate	Moderate

Large	Large
-------	-------

headCircumference

(Genomics England Data Set)

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure centimeters

heartRhythm

(NHIC ICU)

Data Type heartRhythm

Enumeration

Code	Description
19	atrial-ventricular disassociation
17	atrial paced rhythm
18	atrial tachycardia
15	asystole
16	pulseless electrical activity

13	A-V paced rhythm
14	idioventricular rhythm
11	AV block, 3rd degree
12	complete heart block
21	Lt bundle branch block
20	Rt bundle branch block
22	bundle branch block
23	ectopic atrial bradycardia
24	junctional ectopic tachycardia
25	junctional tachycardia
26	paced rhythm
27	sick sinus syndrome
28	supra-ventricular tachycardia
29	SVT with aberrant conduction
3	sinus bradycardia
2	sinus tachycardia
10	AV block, 2nd degree/Wenkebach

1	sinus rhythm
30	ventricular fibrillation
7	wandering atrial pacemaker
6	atrial flutter
5	atrial fibrillation
31	free text
4	sinus arrhythmia
9	AV block, 2nd degree/Mobitz 2
8	AV block, 1st degree

highRVleads

(Genomics England Data Set)

Data Type yesNoUnk

Enumeration

Code	Description
yes	Yes
no	No

unknown	Unknown
---------	---------

holterMonitor

(Genomics England Data Set)

Abnormalities present on the Holter Monitor

Data Type holterAbnormality (Abnormalities present on the Holter monitor)

Enumeration

Code	Description
Atrial fibrillation	Atrial fibrillation
Non-sustained VT	Non-sustained VT
Nil	Nil
Unknown	Unknown

hpoPhenotypes

(Genomics England Data Set)

The identity of the statement within the Human Phenotype Ontology

Data Type xs:string (Character strings in XML.)

Regular Expression HP:[0-9]{7}

imagingModalityFacial

(Annex C)

Imaging Modality Facial

Data Type imagingModalityFacial (Imaging Modality Facial

Enumeration

Code	Description
Medical Photograph	Medical Photograph

imagingProcedure

(Genomics England Data Set)

Based on Modality categories derived from submitted NICIP or SNOMED-CT codes. The following category descriptions have been defined:

Data Type imagingProcedure

Enumeration

Code	Description
------	-------------

Computerised Axial Tomography	Computerized axial tomography
Diagnostic Ultrasonography	Diagnostic ultrasonography (this includes Echocardiogram) (procedure)
Doppler Diastolic Function Assessment	Doppler Diastolic Function Assessment
Endoscopy	Endoscopy
Fluoroscopy	Fluoroscopy
Fundus imaging – Drusen staining by fluorescein angiography	Fundus imaging – Drusen staining by fluorescein angiography
High Speed Video Microscopy	High Speed Video Microscopy
Indocyanin green [ICG] angiography	Indocyanin green [ICG] angiography
Light microscopy	Light microscopy
Magnetic Resonance Imaging	Magnetic Resonance Imaging
Medical Photograph	Photography of patient (procedure)
Nuclear Medicine	Nuclear Medicine
Plain Radiography	Plain radiography (X-ray) (procedure)
Positron Emission Tomography	Positron emission tomography (procedure)
Single Photon Emission Computed Tomography	Single photon emission computed tomography of kidney using dimercaptosuccinic acid (procedure)
SAP scan	SAP scan

Transmission electron microscopy	Transmission electron microscopy
Other	Other

imagingSubModality

(Genomics England Data Set)

Imaging submodalities

Data Type imagingSubModality (Sub-modalities of imaging technologies)

Enumeration

Code	Description
Adrenal imaging	Adrenal imaging
Cardiac MRI	Cardiac Magnetic Resonance Imaging
DEXA	dual energy X-ray absorptiometry
DMSA	dimercaptosuccinic acid scan
Echocardiography	Echocardiography
Ferriscan	Ferriscan
High resolution chest CT	High resolution chest CT
Magnetic Resonance Angiogram	Magnetic Resonance Angiogram

Micturating cystourethrogram	Micturating cystourethrogram
Retinal photography	Retinal photography
Skeletal survey	Skeletal survey
T2*MRI	T2*MRI

infectionDuration

(Genomics England Data Set)

Data Type Infection_Duration

Enumeration

Code	Description
None	None
<7 days	<7 dats
7-13 days	7-13 days
14-42 days	14-42 days
>42 days	>42 days
Unknown	Unknown
No Response	No Response

infectionFrequency

(Genomics England Data Set)

Data Type Infection_Frequency

Enumeration

Code	Description
0-1 per year	0-1 per year
2-4 per year	2-4 per year
>4 per year	>4 per year
Unknown	Unknown
No Response	No Response

interventricularDelay

(Genomics England Data Set)

Data Type yesNoUnk

Enumeration

Code	Description
------	-------------

yes	Yes
no	No
unknown	Unknown

intraocularPressure

(Genomics England Data Set)

Pressure inside the eye measured in mmHg

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure Millimetres of mercury

kidneyRegion

(Annex C)

kidney region

Data Type kidneyRegion

Enumeration

Code	Description
Abdominal	Abdominal

kidneySystem

(Annex C)

kidney system

Data Type kidneySystem (kidney system

Enumeration

Code	Description
Genitourinary	Genitourinary

labAnalysisType

(Genomics England Data Set)

The laboratory analytical test used

Data Type labAnalysisType (enumerated list of laboratory techniques

Enumeration

Codey	Description
Amino acid chromatography	Amino acid chromatography
Arterial blood gas	Arterial blood gas

Auto Antibodies	Auto Antibodies
Bone profile	Bone profile
Cerebrospinal fluid test	Cerebrospinal fluid test
Coeliac screen	Coeliac screen
Comprehensive Metabolic Panel	Comprehensive Metabolic Panel (CMP)
Full Blood Count	Full Blood Count
Liver biochemistry	Liver biochemistry
Microbiology Culture	Microbiology Culture
Muscle enzymes	Muscle enzymes
Pancreatic autoantibodies	Pancreatic autoantibodies
Renal biochemistry	Renal biochemistry
Serum immunoglobulins	Serum immunoglobulins
Sweat chloride test	Sweat chloride test
TORCH screen	TORCH screen
Urine dip	Urine dip

labAnalysisTypeArterialbloodgas

Data Type

labAnalysisTypeArterialbloodgas

Enumeration

Code	Description
Arterial blood gas	Arterial blood gas

labAnalysisTypeAutoantibodies

Data Type labAnalysisTypeAutoantibodies

Enumeration

Codey	Description
Autoantibodies	Autoantibodies

labAnalysisTypeBiotinidase

Data Type labAnalysisTypeBiotinidase

Enumeration

Codeyy	Description
Biotinidase	Biotinidase

labAnalysisTypeBloodTests

Data Type

labAnalysisTypeBloodTests

Enumeration

Codey	Description
Blood Tests	Blood Tests

labAnalysisTypeBoneprofile

Data Type

labAnalysisTypeBoneprofile

Enumeration

Code	Description
Bone profile	Bone profile

labAnalysisTypeCSFtests

Data Type

labAnalysisTypeCSFtests

Enumeration

Code	Description
CSF tests	CSF tests

labAnalysisTypeClotting

Data Type labAnalysisTypeClotting

Enumeration

Code	Description
Clotting	Clotting

labAnalysisTypeCoeliacantibodies

Data Type labAnalysisTypeCoeliacantibodies

Enumeration

Code	Description
Coeliac antibodies	Coeliac antibodies

labAnalysisTypeComplement

Data Type labAnalysisTypeComplement

Enumeration

Code	Description
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Complement	Complement
------------	------------

labAnalysisTypeCongenitalMyaesthesiaAntibodies

Data Type labAnalysisTypeCongenitalMyaesthesiaAntibodies

Enumeration

Code	Description
Congenital Myaesthesia Antibodies	Congenital Myaesthesia Antibodies

labAnalysisTypeCortisol

Data Type labAnalysisTypeCortisol

Enumeration

Code	Description
Cortisol	Cortisol

labAnalysisTypeCryoglobulins

Data Type labAnalysisTypeCryoglobulins

Enumeration

Code	Description
Cryoglobulins	Cryoglobulins

labAnalysisTypeCulture

Data Type labAnalysisTypeCulture

Enumeration

Code	Description
Culture	Culture

labAnalysisTypeExtendedhaematologyinvestigations

Data Type labAnalysisTypeExtendedhaematologyinvestigations

Enumeration

Code	Description
Extended haematology investigations	Extended haematology investigations

labAnalysisTypeExtendedrenalbiochemistry

Data Type labAnalysisTypeExtendedrenalbiochemistry

Enumeration

Code	Description
Extended renal biochemistry	Extended renal biochemistry

labAnalysisTypeFecaltest

Data Type labAnalysisTypeFecaltest

Enumeration

Code	Description
Fecal test	Fecal test

labAnalysisTypeFullBloodCount

Data Type labAnalysisTypeFullBloodCount

Enumeration

Code	Description
Full Blood Count	Full Blood Count

labAnalysisTypeGlucose

Data Type labAnalysisTypeGlucose

Enumeration

Code	Description
Glucose	Glucose

labAnalysisTypeGrowthhormones

Data Type labAnalysisTypeGrowthhormones

Enumeration

Code	Description
Growth hormones	Growth hormones

labAnalysisTypeHormones(other)

Data Type labAnalysisTypeHormones(other)

Enumeration

Code	Description
Hormones (other)	Hormones (other)

labAnalysisTypeInflammatorymarkers

Data Type labAnalysisTypeInflammatorymarkers

Enumeration

Code	Description
Inflammatory markers	Inflammatory markers

labAnalysisTypeInsulinandC-peptide

Data Type labAnalysisTypeInsulinandC-peptide

Enumeration

Code	Description
Insulin and C-peptide	Insulin and C-peptide

labAnalysisTypeLipids

Data Type labAnalysisTypeLipids

Enumeration

Code	Description
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Lipids	Lipids
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labAnalysisTypeLiverbiochemistry

Data Type labAnalysisTypeLiverbiochemistry

Enumeration

Code	Description
Liver biochemistry	Liver biochemistry

labAnalysisTypeMetabolicbiochemistry

Data Type labAnalysisTypeMetabolicbiochemistry

Enumeration

Code	Description
Metabolic biochemistry	Metabolic biochemistry

labAnalysisTypeMicrobiologyantibodies

Data Type labAnalysisTypeMicrobiologyantibodies

Enumeration

Code	Description
Microbiology antibodies	Microbiology antibodies

labAnalysisTypeOtherenzymes

Data Type labAnalysisTypeOtherenzymes

Enumeration

Code	Description
Other enzymes	Other enzymes

labAnalysisTypePancreaticautoantibodies

Data Type labAnalysisTypePancreaticautoantibodies

Enumeration

Code	Description
Pancreatic autoantibodies	Pancreatic autoantibodies

labAnalysisTypePorphyriainvestigations

Data Type labAnalysisTypePorphyriainvestigations

Enumeration

Code	Description
Porphyria investigations	Porphyria investigations

labAnalysisTypePrimaryimmunodeficiencyinvestigations

Data Type labAnalysisTypePrimaryimmunodeficiencyinvestigations

Enumeration

Code	Description
Primary immunodeficiency investigations	Primary immunodeficiency investigations

labAnalysisTypeRenalbiochemistry

Data Type labAnalysisTypeRenalbiochemistry

Enumeration

Code	Description
Renal biochemistry	Renal biochemistry

labAnalysisTypeReninandaldosterone

Data Type labAnalysisTypeReninandaldosterone

Enumeration

Code	Description
Renin and aldosterone	Renin and aldosterone

labAnalysisTypeSerumimmunoglobulins

Data Type labAnalysisTypeSerumimmunoglobulins

Enumeration

Code	Description
Serum immunoglobulins	Serum immunoglobulins

labAnalysisTypeSexhormones

Data Type labAnalysisTypeSexhormones

Enumeration

Code	Description
Sex hormones	Sex hormones

labAnalysisTypeTORCHscreen

Data Type labAnalysisTypeTORCHscreen

Enumeration

Code	Description
TORCH screen	TORCH screen

labAnalysisTypeThyroidfunctiontesting

Data Type labAnalysisTypeThyroidfunctiontesting

Enumeration

Code	Description
Thyroid function testing	Thyroid function testing

labAnalysisTypeUrineDip

Data Type labAnalysisTypeUrineDip

Enumeration

Code	Description
------	-------------

Urine Dip	Urine Dip
-----------	-----------

labAnalysisTypeUrineTest

Data Type labAnalysisTypeUrineTest

Enumeration

Code	Description
Urine Test	Urine Test

labAnalysisTypeUrinedip-standard

Data Type labAnalysisTypeUrinedip-standard

Enumeration

Code	Description
Urine dip - standard	Urine dip - standard

labAnalysisTypeUrinedip-sulphites

Data Type labAnalysisTypeUrinedip-sulphites

Enumeration

Code	Description
Urine dip - sulphites	Urine dip - sulphites

labAnalysisTypeUrineelectrolytes

Data Type labAnalysisTypeUrineelectrolytes

Enumeration

Code	Description
Urine electrolytes	Urine electrolytes

labAnalysisTypeUrineglucose

Data Type labAnalysisTypeUrineglucose

Enumeration

Code	Description
Urine glucose	Urine glucose

labAnalysisTypeUrinemetabolictests

Data Type labAnalysisTypeUrinemetabolictests

Enumeration

Code	Description
Urine metabolic tests	Urine metabolic tests

labAnalysisTypeUrineprotein

Data Type labAnalysisTypeUrineprotein

Enumeration

Code	Description
Urine protein	Urine protein

labAnalysisTypeUrinereducingsubstances

Data Type labAnalysisTypeUrinereducingsubstances

Enumeration

Code	Description
Urine reducing substances	Urine reducing substances

labAnalysisTypeVirology

Data Type labAnalysisTypeVirology

Enumeration

Code	Description
Virology	Virology

labAnalysisTypeVitaminB12

Data Type labAnalysisTypeVitaminB12

Enumeration

Code	Description
Vitamin B12	Vitamin B12

laboratorySampleId

(Genomics England Data Set)

Laboratory Sample ID (GS1 Data Matrix)

Data Type xs:string (Character strings in XML.

lengthInCm

(Genomics England Data Set)

Data Type decimal ((cm)

Unit of Measure centimeters

lengthInMm

(Genomics England Data Set)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E12.78e-2, 12 and INF

Unit of Measure Millimetres

leucocyteDipResult

(Genomics England Data Set)

Data Type leucocyteDipResult

Enumeration

Code	Description
Negative	Negative
Trace	Trace
Small	Small
Moderate	Moderate

Large	Large
-------	-------

lifeStatus

Data Type lifeStatus

Enumeration

Code	Description
alive	alive
aborted	aborted
deceased	deceased
unborn	unborn
stillborn	stillborn
miscarriage	miscarriage

liverRegion

(Annex C)

Liver Region

Data Type liverRegion

liverSystem

(Annex C)

liver system

Data Type liverSystem

localSampleId

(Rare Diseases)

The local identifier for the source sample

Data Type xs:string (Character strings in XML.

lowMolecularWeightProtein

(Genomics England Data Set)

Data Type highLowNormal

Enumeration

Code	Description
High	Raised above normal level
Low	Reduced below normal level

Normal	At a normal level
--------	-------------------

mass

(Genomics England Data Set)

mass in kg

Data Type	<p>xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF</p>
Unit of Measure	<p>kilogram (The international prototype of the kilogram, an artefact made of platinum-iridium, is kept at the BIPM under the conditions specified by the 1st CGPM in 1889 when it sanctioned the prototype and declared:</p> <p>This prototype shall henceforth be considered to be the unit of mass. The 3rd CGPM (1901), in a declaration intended to end the ambiguity in popular usage concerning the use of the word "weight", confirmed that:</p> <p>The kilogram is the unit of mass; it is equal to the mass of the international prototype of the kilogram. The complete declaration appears here.</p> <p>It follows that the mass of the international prototype of the kilogram is always 1 kilogram exactly, $m(\text{grand K}) = 1 \text{ kg}$. However, due to the inevitable accumulation of contaminants on surfaces, the international prototype is subject to reversible surface contamination that approaches 1 μg per year in mass. For this reason, the CIPM declared that, pending further research, the reference mass of the international prototype is that immediately after cleaning and washing by a specified method (PV, 1989, 57, 104-105 and PV, 1990, 58, 95-97). The reference mass thus defined is used to calibrate national standards of platinum-iridium alloy (Metrologia, 1994, 31, 317-336).</p>

nhsNumber

(NHS Data Dictionary)

[Http://www.datadictionary.nhs.uk/data_dictionary/attributes/n/nhs/nhs_number_de.asp?query=nhs%20number&rank=100&shownav=1](http://www.datadictionary.nhs.uk/data_dictionary/attributes/n/nhs/nhs_number_de.asp?query=nhs%20number&rank=100&shownav=1)

The NHS NUMBER, the primary identifier of a PERSON, is a unique identifier for a PATIENT within the NHS in England and Wales. This will not vary by any ORGANISATION of which a PERSON is a PATIENT. It is mandatory to record the NHS NUMBER. There are exceptions, such as Accident and Emergency care, sexual health and major incidents, as defined in existing national policies.

The NHS NUMBER is 10 numeric digits in length. The tenth digit is a check digit used to confirm its validity. The check digit is validated using the Modulus 11 algorithm and the use of this algorithm is mandatory. There are 5 steps in the validation of the check digit. Further guidance is available from the Health and Social Care Information Centre website.

Data Type xs:string (Character strings in XML.

Rule

```

def isValid = false
if (x.size() == 10) {
  Integer total = 0
  Integer i = 0
  for (i = 0; i <= 8; i++) {
    def digit = x.substring(i, (i+1))
    def factor = 10 - i
    total = total + (digit.toInteger() * factor) }
  def checkDigit = (11 - (total.mod(11)))
  if (checkDigit == 11) { checkDigit = 0 }
  def check = x.substring(9,10)
  if (check.toInteger() == checkDigit && checkDigit!=10) { isValid = true }
}
return isValid

```

non-imagingMethods

(Genomics England Data Set)

Data Type non-imagingMethods (Types of non-imaging diagnostic techniques

Enumeration

Code	Description
------	-------------

Auditory Brainstem Response	Auditory Brainstem Response
Cardiac drug challenge test	Cardiac drug challenge test
Electrocardiography	Electrocardiography
Electrophysiological study	Electrophysiological study
Electroencephalogram	Electroencephalogram
Electromyography	Electromyography
Electroretinography	Electroretinography
Electro-oculography	Electro-oculography
Exercise test	Exercise test
Hearing test	Hearing test
Holter monitor	Holter monitor
Ishihara color plates	Ishihara color plates
Keratometry	Keratometry
Lung function tests	Lung function tests
Motor nerve conduction studies	Motor nerve conduction studies
Motor evoked potentials	Motor evoked potentials
Multiple sleep latency test	Multiple sleep latency test

Nerve conduction studies	Nerve conduction studies
Otoacoustic Emissions	Otoacoustic Emissions
Phototest response to UVA	Phototest response to UVA
Polysomnography	Polysomnography
Repetitive nerve stimulation	Repetitive nerve stimulation
Sensory nerve conduction studies	Sensory nerve conduction studies
Stone analysis	Stone analysis
Visual evoked potential study	Visual evoked potential study
Water deprivation test	Water deprivation test
Other	Other

nonImagingMethodAuditoryBrainstemResponse

Data Type nonImagingMethodAuditoryBrainstemResponse

Enumeration

Code	Description
Auditory Brainstem Response	Auditory Brainstem Response

nonImagingMethodColourPlateTest

Data Type nonImagingMethodColourPlateTest

Enumeration

Code	Description
Colour Plate Test	Colour Plate Test

nonImagingMethodDarkAdaptationTest

Data Type nonImagingMethodDarkAdaptationTest

Enumeration

Code	Description
Dark Adaptation Test	Dark Adaptation Test

nonImagingMethodECGdiagnostics

Data Type nonImagingMethodECGdiagnostics

Enumeration

Code	Description
ECG diagnostics	ECG diagnostics

nonImagingMethodElectro-oculogram

Data Type nonImagingMethodElectro-oculogram

Enumeration

Code	Description
Electro-oculogram	Electro-oculogram

nonImagingMethodElectrophysiologicalstudy

Data Type nonImagingMethodElectrophysiologicalstudy

Enumeration

Code	Description
Electrophysiological study	Electrophysiological study

nonImagingMethodElectroretinogram

Data Type nonImagingMethodElectroretinogram

Enumeration

Code	Description
------	-------------

Electroretinogram	Electroretinogram
-------------------	-------------------

nonImagingMethodEpinephrinechallenge

Data Type nonImagingMethodEpinephrinechallenge

Enumeration

Code	Description
Epinephrine challenge	Epinephrine challenge

nonImagingMethodExercisetest-cardiac

Data Type nonImagingMethodExercisetest-cardiac

Enumeration

Code	Description
Exercise test - cardiac	Exercise test - cardiac

nonImagingMethodForcedvitalcapacity

Data Type nonImagingMethodForcedvitalcapacity

Enumeration

Code	Description
Forced vital capacity	Forced vital capacity

nonImagingMethodHeartobservations

Data Type nonImagingMethodHeartobservations

Enumeration

Code	Description
Heart observations	Heart observations

nonImagingMethodHoltermonitortest

Data Type nonImagingMethodHoltermonitortest

Enumeration

Code	Description
Holter monitor test	Holter monitor test

nonImagingMethodOcularMalformationMetrics

Data Type nonImagingMethodOcularMalformationMetrics

Enumeration

Code	Description
Ocular Malformation Metrics	Ocular Malformation Metrics

nonImagingMethodOcularPressure

Data Type nonImagingMethodOcularPressure

Enumeration

Code	Description
Ocular Pressure	Ocular Pressure

nonImagingMethodOtoacousticEmissions

Data Type nonImagingMethodOtoacousticEmissions

Enumeration

Code	Description
Otoacoustic Emissions	Otoacoustic Emissions

nonImagingMethodRefractionError

Data Type nonImagingMethodRefractionError

Enumeration

Code	Description
Refraction Error	Refraction Error

nonImagingMethodSignalaveragedECG

Data Type nonImagingMethodSignalaveragedECG

Enumeration

Code	Description
Signal averaged ECG	Signal averaged ECG

nonImagingMethodSleeptest

Data Type nonImagingMethodSleeptest

Enumeration

Code	Description
Sleep test	Sleep test

nonImagingMethodSodiumchannelblockerchallenge

Data Type nonImagingMethodSodiumchannelblockerchallenge

Enumeration

Code	Description
Sodium channel blocker challenge	Sodium channel blocker challenge

nonImagingMethodVisualAcuity

Data Type nonImagingMethodVisualAcuity

Enumeration

Code	Description
Visual Acuity	Visual Acuity

nonImagingMethodVisualField

Data Type nonImagingMethodVisualField

Enumeration

Code	Description
------	-------------

Visual Field	Visual Field
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normalElevated

(Genomics England Data Set)

Data Type normalElevated (Indicating a normal or elevated test result)

Enumeration

Code	Description
Normal	normal
Elevated	elevated
Unknown	unknown

numberUnitOfMeasurement

(Genomics England Data Set)

Data Type number

Enumeration

Code	Description
Number (Retired September 2013)	Number (Retired September 2013)

Percentage (%)	Percentage (%)
----------------	----------------

organisationCode

(NHS Data Dictionary)

An Organisation code or ODS code identifies an NHS Organisation uniquely

Data Type xs:string (Character strings in XML.

otoacousticEmissionsResult

(Genomics England Data Set)

Data Type otoacousticEmissionsResult (Result of OAE test

Enumeration

Code	Description
Present	Present
Absent	Absent
Disappeared but Cochlear microphonic still present	Disappeared but Cochlear microphonic still present
Disappeared and cochlear microphonic absent	Disappeared and cochlear microphonic absent

participantId

Genomics England participant identifier (supplied by Genomics England)

Data Type xs:string (Character strings in XML.

Rule maxLength(12) && minLength(3)

participantType

(Genomics England Data Set)

The participant type in a medical genetic study i.e. whether the person is a proband or a relative

Data Type ParticipantType (The participant type in a medical genetic study i.e. whether the person is a proband or a relative

Enumeration

Code	Description
Proband	Proband
Relative	Relative

patientStatus

(Genomics England Data Set)

Represents the patient's status when test was performed

Data Type patientStatus

Enumeration

Code	Description
presenting	Taken at patient presentation
diagnostic	Taken at the point of diagnosis
baseline	Representing a baseline measurement
most abnormal	Representing the most abnormal measurement
unknown	Status unknown

patternOfLvHypertrophy

(Genomics England Data Set)

Data Type HypertrophyPattern (Pattern of LV hypertrophy)

Enumeration

Code	Description
Symmetric	Symmetric
Asymmetric (maximum wall thick/min wall thick of >1.3)	Asymmetric (maximum wall thick/min wall thick of >1.3)
Unknown	Unknown

penetrance

(Genomics England Data Set)

Data Type yesNoUnk (Yes / no / unknown)

Enumeration

Code	Description
N	No
9	Unknown
Y	Yes

personFamilyName

(Cancer Outcomes and Services Dataset)

That part of a PERSON's name which is used to describe family, clan, tribal group, or marital association.

Data Type xs:string (Character strings in XML.

Rule maxLength(35)

personFamilyName(atBirth)

(Cancer Outcomes and Services Dataset)

The PATIENT's surname at birth.

Data Type	xs:string (Character strings in XML.
Rule	maxLength(35)

personGivenName

(Cancer Outcomes and Services Dataset)

The forename(s) or given name(s) of a PERSON.

Data Type	xs:string (Character strings in XML.
Rule	maxLength(35)

personHeightInMetres

(Cancer Outcomes and Services Dataset)

Height of the patient, in metres, to 2 decimal places (n.nn).

Data Type	xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF
Unit of Measure	meters

personKaryotypicSexClassification

(Genomics England Data Set)

Data Type	personKaryotypicSex
-----------	---------------------

Enumeration

Code	Description
XY	XY
XX	XX
XO	XO
XXY	XXY
XXY	XXY
XXX	XXX
XXYY	XXYY
XXXY	XXXY
XXXX	XXXX
other	other
unknown	unknown

personObservation(weight)

(Cancer Outcomes and Services Dataset)

Weight of the patient, in kilograms with up to three decimal places (nnn.nnn).

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN})

Example, -1E4, 12.78e-2, 12 and INF

kilogram (The international prototype of the kilogram, an artefact made of platinum-iridium, is kept at the BIPM under the conditions specified by the 1st CGPM in 1889 when it sanctioned the prototype and declared:

This prototype shall henceforth be considered to be the unit of mass.

The 3rd CGPM (1901), in a declaration intended to end the ambiguity in popular usage concerning the use of the word "weight", confirmed that:

Unit of Measure

The kilogram is the unit of mass; it is equal to the mass of the international prototype of the kilogram.

The complete declaration appears here.

It follows that the mass of the international prototype of the kilogram is always 1 kilogram exactly, $m(\text{grand K}) = 1 \text{ kg}$. However, due to the inevitable accumulation of contaminants on surfaces, the international prototype is subject to reversible surface contamination that approaches $1 \mu\text{g}$ per year in mass. For this reason, the CIPM declared that, pending further research, the reference mass of the international prototype is that immediately after cleaning and washing by a specified method (PV, 1989, 57, 104-105 and PV, 1990, 58, 95-97). The reference mass thus defined is used to calibrate national standards of platinum-iridium alloy (Metrologia, 1994, 31, 317-336).

personPhenotypicSexClassification

(NHS Data Dictionary)

A classification of PERSON PHENOTYPIC SEX

http://www.datadictionary.nhs.uk/data_dictionary/attributes/p/person/person_phenotypic_sex_classification_de.asp?shownav=1

Data Type

personPhenotypicSex (A classification of PERSON PHENOTYPIC SEX)

Enumeration

Code	Description
2	Female

1	Male
9	Indeterminate

personStatedGenderCode

(Cancer Outcomes and Services Dataset)

Person's gender as self-declared (or inferred by observation for those unable to declare their PERSON STATED GENDER).

Data Type personStatedGenderCode (Person's gender as self-declared (or inferred by observation for those unable to declare their PERSON STATED GENDER)).

Enumeration

Code	Description
2	Female
1	Male
9	Not Specified (Unable to be classified as either male or female)
X	Not Known (PERSON STATED GENDER CODE not recorded)

positiveInteger

(Genomics England Data Set)

Data Type	xs:nonNegativeInteger (Infinite set {0, 1, 2,...}. Sign omitted, “+” assumed. Example: 1, 0, 12678967543233, +100000.
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Rule	minInclusive(0)
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pressureInMmHg

(Genomics England Data Set)

Data Type	xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.
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Unit of Measure	Millimetres of mercury
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procedure(snomedCt)

(Cancer Outcomes and Services Dataset)

snomed ct procedures

Data Type	snomedCt
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procedureCodeSnomedCT

(SNOMED CT)

snomed ct procedure code subset

Data Type	snomedCt
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proteinDeposition

(Genomics England Data Set)

Data Type

proteinDeposition (Values for protein deposition result from electron microscopy testing in renal biopses)

Enumeration

Code	Description
amyloid	Amyloid
fibrin	Fibrin
immunotactoid	Immunotactoid
other	Other

rackId

(Genomics England Data Set)

Barcode on the containing rack as dispatched (128 Barcode)

Data Type

xs:string (Character strings in XML.)

rackWell

(Genomics England Data Set)

The GMC must record the position of the sample in the Rack they send to the biorepository. Each rack is has 96 wells. The position of a sample in these wells is coded from A-H and 1-12

Data Type xs:string (Character strings in XML.

Regular Expression [A-H][1-2] | [1-9]

sampleId

(Genomics England Data Set)

The sample id i.e the barcode from the sample tube used for sample collection at the GMC clinic.
This will be a Code-128 Barcode

Data Type xs:string (Character strings in XML.

samplingPreconditions

(Rare Diseases)

Data Type samplingPreconditions

Enumeration

Code	Description
Fasting sample	Post Fasting Sample
Post mortem sample	Post Mortem Sample
Post-dose sample	Post-dose sample
Pre-dose sample	Pre-dose sample

Random Sample	Random Sample
Sample from ambulatory subject	Sample from ambulatory subject
Sample from orthostatic subject	Sample from orthostatic subject
Sample from rested subject	Sample from rested subject
Sample from subject of unknown posture	Sample from subject of unknown posture
Sample from supine subject	Sample from supine subject

smokingStatus

(Cancer Outcomes and Services Dataset)

Specify the current smoking status of the patient.

Data Type smokingStatus (Specify the current smoking status of the patient.)

Enumeration

Code	Description
1	Current smoker
2	Ex smoker
3	Non-smoker - history unknown
4	Never smoked

Z	Not Stated (PERSON asked but declined to provide a response)
9	Unknown

snomedCtDm+d

(Rare Diseases)

[Http://www.dmd.nhs.uk/snomed_links/index.html](http://www.dmd.nhs.uk/snomed_links/index.html)

<https://isd.hscic.gov.uk/trud3/user/guest/group/0/pack/6>

Data Type xs:string (Character strings in XML.

sodiumChannelBlocker

(Genomics England Data Set)

The sodium channel blocker used in a drug challenge test

Data Type sodiumChannelBlocker (The Na channel blocker used in a drug challenge test

Enumeration

Code	Description
ajmanline	Ajmanline
flecainide	Flecainide

sodiumChannelBlockerDosage

(Genomics England Data Set)

The dosage of a sodium channel blocker used in a drug challenge test

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure milligrams per kilogram (A drug dosage per kg of body weight

sourceOrganisation

(Genomics England Data Set)

source organisation

Data Type sourceOrganisation (Source organisation

Enumeration

Code	Description
GMC	Genomic Medicine Centre
BIO	Biorepository
SEQ	Sequencer

spatialPattern

(Human Phenotype Ontology)

The pattern by which a phenotype affects one or more regions of the body.

Data Type spatialPattern (The pattern by which a phenotype affects one or more regions of the body.

Enumeration

Code	Description
Distal	Localized away from the central point of the body.
Generalized	Affecting all regions without specificity of distribution.
Localized	Being confined or restricted to a particular location.
Proximal	The pattern by which a phenotype affects one or more regions of the body.

specialistRenalUnitOfMeasurement

(Genomics England Data Set)

Data Type specialistRenal

Enumeration

Code	Description
Litres per week per 1.73 metres squared (l/week/1.73 ²)	Litres per week per 1.73 metres squared (l/week/1.73 ²)
Microgram albumin per 24 hours (µg/24hr)	Microgram albumin per 24 hours (µg/24hr)

Microgram albumin per hour ($\mu\text{g/ml/hr}$)	Microgram albumin per hour ($\mu\text{g/ml/hr}$)
Microgram albumin per minute ($\mu\text{g/min}$)	Microgram albumin per minute ($\mu\text{g/min}$)
Millilitres per Minute divided by 1.73 Square Metres ($\text{ml/min}/1.73\text{m}^2$)	Millilitres per Minute divided by 1.73 Square Metres ($\text{ml/min}/1.73\text{m}^2$)

stimulation

(Genomics England Data Set)

Data Type stimulationType

Enumeration

Code	Description
flicker	Flicker stimulation
mixed/combined	Mixed/Combined stimulation

testResult

lab test result

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

thicknessInMicrometres

(Genomics England Data Set)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Micrometre

thicknessInMm

(Genomics England Data Set)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Millimetres

timeAspect

(Rare Diseases)

Data Type timeAspect

Enumeration

Code	Description
Spot	Spot
Unspecified	Unspecified
24hr Collection	24hr Collection

Basal	Basal - Time Course
Stimulated Peak	Stimulated Peak - Time Course

timeInMs

(Genomics England Data Set)

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure Millisecond

timeIntervalMs

(Genomics England Data Set)

time interval in milliseconds

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Millisecond

tissueOrigin

(Annex C)

Origin of the tissue

Data Type

tissueOrigin (The tissue of origin of the sample

Enumeration

Code	Description
Blood	Blood
Skin	Skin
Saliva	Saliva
Muscle	Muscle
CVS	CVS
Amniocentesis	Amniocentesis
Tumour	Tumour
Other	Other

type1ECG

(Genomics England Data Set)

Data Type

yesNoUnk

Enumeration

Code	Description
yes	Yes
no	No
unknown	Unknown

typeOfMeasurement

(Genomics England Forms)

Data Type

Type_of_measurement

Enumeration

Code	Description
PatientReported	Patient reported
Measured	Measured
Unknown	Unknown

verp

(Genomics England Data Set)

VERP at S1 drive train 600ms

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure Millisecond

visualAcuity

(Genomics England Data Set)

Data Type visualAcuity

Enumeration

Code	Description
20/_	20/_
CF	CF
HM	HM
LP	LP
NLP	NLP

visualAnalogPainScore

(Genomics England Data Set)

Data Type xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN}
Example, -1E4, 12.78e-2, 12 and INF

Unit of Measure centimeters

volumelnMicroliters

(SI)

Volume in microliters

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure microliters (Volume in microliters

volumelnMl

(Genomics England Data Set)

Data Type xs:decimal (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Unit of Measure millilitres

volumeUnitOfMeasurement

(Genomics England Data Set)

Data Type volume

Enumeration

Code	Description
Cubic Millimetres (mm3)	Cubic Millimetres (mm3)
Decilitres (dl)	Decilitres (dl)
Femtolitres (fl)	Femtolitres (fl)
Litres (l)	Litres (l)
Millilitres (ml)	Millilitres (ml)
Millilitres (ml) (Retired September 2013)	Millilitres (ml) (Retired September 2013)
Nanograms per litre (ng/l)	Nanograms per litre (ng/l)

xs:base64Binary

(XMLSchema)

Base64-encoded arbitrary binary data

Data Type xs:base64Binary (Base64-encoded arbitrary binary data)

Regular Expression [a-zA-Z0-9=]*

xs:date

(XMLSchema)

Calendar date.Format YYYY-MM-DD. Example, May the 31st, 1999 is: 1999-05-31.

Data Type `xs:date` (Calendar date.Format YYYY-MM-DD. Example, May the 31st, 1999 is: 1999-05-31.

Rule `import static javax.xml.bind.DatatypeConverter.*`

`parseDateTime(string(x))` in Calendar

xs:dateTime

(XMLSchema)

Specific instant of time. ISO 8601 extended format YYYY-MM-DDThh:mm:ss. Example, to indicate 1:20 pm on May the 31st, 1999 for Eastern Standard Time which is 5 hours behind Coordinated Universal Time (UTC): 1999-05-31T13:20:00-05:00.

Data Type `xs:dateTime` (Specific instant of time. ISO 8601 extended format YYYY-MM-DDThh:mm:ss. Example, to indicate 1:20 pm on May the 31st, 1999 for Eastern Standard Time which is 5 hours behind Coordinated Universal Time (UTC): 1999-05-31T13:20:00-05:00.

Rule `import static javax.xml.bind.DatatypeConverter.*`

`parseDateTime(string(x))` in Calendar

xs:decimal

(XMLSchema)

Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Data Type `xs:decimal` (Arbitrary precision decimal numbers. Sign omitted, “+” is assumed. Leading and trailing zeroes are optional. If the fractional part is zero, the period and following zero(es) can be omitted.

Rule `import static javax.xml.bind.DatatypeConverter.*`

`parseDecimal(string(x))` in `BigDecimal`

xs:double

(XMLSchema)

Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF

Data Type	xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF
Rule	<pre>import static javax.xml.bind.DatatypeConverter.* parseDouble(string(x)) in Double</pre>

xs:integer

(XMLSchema)

Integer or whole numbers - Sign omitted, “+” is assumed. Example: -1, 0, 12678967543233, +100000

Data Type	xs:integer (Integer or whole numbers - Sign omitted, “+” is assumed. Example: -1, 0, 12678967543233, +100000
Rule	<pre>import static javax.xml.bind.DatatypeConverter.* parseInteger(string(x)) in BigInteger</pre>

xs:nonNegativeInteger

(XMLSchema)

Infinite set {0, 1, 2,...}. Sign omitted, “+” assumed. Example: 1, 0, 12678967543233, +100000.

Data Type	xs:nonNegativeInteger (Infinite set {0, 1, 2,...}. Sign omitted, “+” assumed. Example: 1, 0, 12678967543233, +100000.
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Rule	minInclusive(0)
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xs:string

(XMLSchema)

Character strings in XML.

Data Type	xs:string (Character strings in XML.
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Rule	import static javax.xml.bind.DatatypeConverter.* true && (x = parseString(string(x)))
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years(fractions)

(SI)

Data Type	xs:double (Double-precision 64-bit floating point type legal literals {0, -0, INF, -INF and NaN} Example, -1E4, 12.78e-2, 12 and INF
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Unit of Measure	Year (A year is the orbital period of the Earth moving in its orbit around the Sun
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yesNo

Boolean, yes no response

Data Type yesNo

Enumeration

Code	Description
yes	Yes
no	No

yesNoUnk

Data Type yesNoUnk

Enumeration

Code	Description
yes	yes
no	no
unknown	unknown

yesnunknown

(Genomics England Data Set)

Data Type yesNoUnk

Enumeration

Code	Description
yes	yes
no	no
unknown	unknown