Genomics England

Rare Disease Model

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Introduction  
  
Purpose  
  
The purpose of this document is to describe the data that Genomic Medicine Centres are asked to supply to accompany the samples submitted for analysis. This document is intended to be read in conjunction with the User Guide and describe the classes of data expected, the association between diseases and the classes of data deemed relevant, and the datatypes employed.   
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  
  
Audience  
  
This document is primarily written for informatics leads within the GMCs and those involved in the collection and submission of data for the UK 100,000 Genomes Project.  
  
Related Documents  
  
This document should be read in conjunction with:  
• Genomics England Data Model Catalogue (Genomics England Model Catalogue)  
• Schemas (XSDs)  
• Example XML Files   
• NHS England GMC Service Specification   
• Relevant Appendices  
  
How to use this document  
  
This document is split into sections that describe the information we expect to receive within the context of each xml submission. The document is primarily split by schema. Within each schema we expect to receive a set of classes of data. Within each class we expect to receive a number of data elements, each of which is associated with a name, a brief explanation, a multiplicity, a datatype and often a business rule.  
NOTE: this document should be used in conjunction with the User Guidance for Cancer Data Specification Document.  
In this version of the document, the data item identifiers have been included, to facilitate look-up in the current version of the on-line metadata catalogue. In addition you can click on the value link to view the full definition for the data type and the applicable constraints.  
SCHEMAS  
Define the classes and data elements included in each xml submission. Each schema describes the information within the corresponding xml document. Each class describes the data elements included within each section of the schema. Each data element has a data type and some have business rules. Click on the hyperlinks in the document to navigate to each of these.  
DATA TYPES:  
Types, rules and enumerations that constrain the value of a data element.  
Rules are expressed as regular expressions and/or groovy code.  
Enumerations are described by their code and description.   
NOTE: Enumerations in grey are deprecated. Although deprecated codes will continue to be accepted they will be removed from the next release and users are encouraged to use other suitable codes.  
BUSINESS RULES:  
Contains business rules that apply across data elements within the context of a file submission.  
  
  
Core data  
  
Clinical information as part of the Core data or Additional data should be collected using several mechanisms:  
• Human phenotype ontology (HPO) data – detailed information about the patient’s symptoms and signs is collected using standardised terms from the HPO. This has been designed to collect information about medical conditions in a way which is easily compared between many individuals in a large dataset  
• ‘Clinical evaluation’ and ‘Clinical test’ data – some clinical information cannot be captured in HPO, for example test results which require a number and unit to be collected. In all participants, basic growth parameters are requested and in all with a disorder of onset under 16 years, information regarding developmental milestones and pregnancy details – this is referred to as ‘Clinical evaluation data’. Additional, pertinent clinical investigations, and imaging data are collected separately in the ‘Clinical Tests’ form.  
• Genetic investigation results – are also collected as part of the Clinical Tests form. These are important to help make diagnoses in patients (e.g. if a patient has a recessive condition and one mutation has already been found, the bioinformaticians will need to compare this result with the genome sequence results), and also to answer questions about how the results of genome sequencing compare with current forms of genetic testing.  
• Pedigree – family history information will be captured in the form of a pedigree diagram, which will be entered using locally-selected pedigree-drawing software then exported in a standardised format for use in the analysis.  
  
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.   
  
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.   
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.   
  
  
Completeness  
  
Essential data items are required on all participants. Core data items are required as set out below. Additional data items are core data items that occur after submission of the initial data set and are required on the same participants as the corresponding core data items.   
  
All participants:  
Essential data set including details of consent and diagnoses  
Updates on changes to consent status and death  
  
All probands:  
A pedigree data set   
  
Affected participants (including probands):  
The core data set including:  
At least one set of ‘general observations’  
At least one phenotyping report\*   
Details of previous genetic investigations  
Clinical test data\*  
Reports of clinical events  
  
Unaffected participants:  
The following components of the core data set:  
At least one set of ‘general observations’  
Phenotyping report(s) if clinically relevant\*\*  
Clinical test data if clinically relevant\*\*\*  
Any other elements of the core data set that are relevant to that participant  
  
\*Phenotype and clinical test data required for each rare disorder are specified in the disease-specific components of the data model.  
\*\*Relevant phenotyping reports in an unaffected participant would include the presence or absence of phenotypes that are relevant to the phenotypes in the family, for example the absence of renal cysts in the parent of a child with cystic renal disease.  
\*\*\*Relevant clinical test data in an unaffected participant would including positive or negative test results relevant to the phenotypes in the family, for example a normal ECG in the parent of a child with long QT syndrome.  
  
From a data modelling perspective most of the classes are ‘optional’, with the exception of the registration and consent information, in that an event of that class may not yet have occurred, or may not yet have been reported, for a particular participant.   
Event records or reports are required for all relevant clinical events or observations to date (for the core data).  
For some classes of event, a report will be accepted only if additional classes are provided as part of the report and/or other 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 or 0..\* it is considered ‘non-mandatory’ and 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.  
  
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 policies 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 finalised.  
  
  
XML Submission  
  
All submissions in XML format must include as metadata: the date and time upon which the XML file was generated; the name and version of the schema used for validation; and the organisation within the GMC responsible for the participant. A source system identifier and a local report identifier may also be included.  
The data provided in XML format must include an event date and event reference for the report in question. This reference should be unique within the GMC. If a second submission is received against the same event reference then this will be treated as an update.

XML Schema (.XSD Files)

The XML files and the Rare Disease Model Data Specification v2 are generated from the Genomics England Data Model Catalogue. The XML schemas can be downloaded directly from the Model Catalogue within the Assets folder for the Model.  
For access to the Rare Disease Data Model Catalogue, and to submit any comments, observations or issues, please contact the Genomics England Contact Desk (see Section 14 in this document regarding support and queries).

XML Validation to take place before submission of file

The use of XML was mandated in the e-Government Interoperability Framework (eGIF) as a messaging standard between government organisations and has been adopted by NHS (http://systems.digital.nhs.uk/data/nhsdmds/ddcn/cr1345.pdf) and therefore, this programme. XML delivers some rigour to messaging by controlling the message structure, and the data element contents and format through an XML schema definition (XSD).   
It is essential that NHS GMCs send files that comply with their corresponding schema. This is done by ‘validation’, an electronic process that compares an XML message against its XSD. There are a number of online tools that provide this service – including free tools such as: Notepad++ (https://notepad-plus-plus.org/). There are also richer paid-for tools that provide a graphical view. The NHS recommends Altova (https://www.altova.com/) and GeL uses oXygen XML (https://www.oxygenxml.com/).   
The Model Catalogue also offers XML validation functionality by selecting ‘validate xml’ from the asset menu.  
All submissions not passing validation will be rejected and an email will be sent advising of the failure and the reason from the Data Acquisition and Management system to any individuals subscribed to warning messages for that GMC.

FAQs  
  
Essential data  
Patient/Ethnicity  
’99 - Unknown’ has been added in the latest release.

# Essential Data

## Registration and Consent

The GMC clinic is expected to establish eligibility, to validate NHS numbers before registration 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.

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Registration and Consent report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### Registration [1..1]

*One report containing Registration must be submitted together with each Registration and Consent report.*

The proband’s participant ID should be used as the family ID for all members of the recruited family unit.  
In gender related questions, please outline the participant’s stated gender and phenotypic sex at birth (if known).   
In the Genomic Centre identifier section, please fill in the ODS code of the NHS trust responsible for the patient.   
Ethnicity data should be captured, as stated by the participant.  
Entry of ’Surname at birth’ is required only if different from current surname.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Family ID (12504)** | Genomics England Family Identifier assigned to the proband and their relatives. This should be the Proband Participant ID. | 1..1 | [gmcFamilyId](#33502) |  |
| **Person Stated Gender (12509)** | The participant's current gender | 1..1 | [personStatedGenderCode](#54281) **1**:Male **2**:Female **9**:Indeterminate (Unable to be classified as either male or female)  **X**:Not Known (PERSON STATED GENDER CODE not recorded) | PERSON STATED GENDER CODE (CR3170 from Cancer Outcomes and Services Dataset) |
| **Person Phenotypic Sex (12510)** | The participant's sex classification at birth. 9 - Indeterminate, may only be used if the patients chromosomal sex at birth is ambiguous. Samples cannot be sequenced until the chromosomal sex is established therefore queries will be raised for all patients submitted with a value of 9 to confirm that chromosomal sex is indeterminate rather than unknown prior to sequencing. | 1..1 | [personPhenotypicSexClassification](#42016) **2**:Female **1**:Male **9**:Indeterminate |  |
| **Person Karyotypic Sex (31267)** | The participant’s karyotypic sex if known and not as expected for gender or phenotypic sex | 1..1 | [personKaryotypicSexClassification](#33949) |  |
| **Fetal Participant (39046)** | State whether this is a fetal participant | 1..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |
| **Surname at Birth (12511)** | The participant's surname at birth, if available and different from current surname | 0..1 | [personFamilyNameAtBirth](#54277) | PERSON FAMILY NAME (AT BIRTH) (CR0140 from Cancer Outcomes and Services Dataset) |
| **Recruiting Trust ID (14860)** | ODS code of the recruiting trust – LDP (Local Delivery Partner) or main GMC trust | 1..1 | [organisationSiteCode](#42015) |  |
| **Ethnicity (14445)** | 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](#54205) | ETHNIC CATEGORY (CR0150 from Cancer Outcomes and Services Dataset) ETHNIC CATEGORY (CR0150 from Cancer Outcomes and Services Dataset) |
| **Local Case Identifier (33982)** | Optional case identifier or family number used locally, if different from family id | 0..1 | [xs:string](#38) |  |

#### Event Details [1..1]

*One report containing Event Details must be submitted together with each Registration report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

#### Participant Contact Details [1..1]

*One report containing Participant Contact Details must be submitted together with each Registration report.*

Please include all available contact details for the participant. At least one set of the contact details field MUST be supplied.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant Email Address (12529)** | Email address of participant | 0..1 | [emailAddress](#54204) |  |
| **Participant Home Telephone (12532)** | If available, the participant's home telephone number | 0..1 | [ukTelephoneNumber](#54308) |  |
| **Participant Mobile Telephone (12533)** | If available, the participant's mobile telephone number | 0..1 | [ukTelephoneNumber](#54308) |  |
| **Address line 1 (12822@0.0.1 )** | Premises ID and/or house name, e.g. 'Flat 1', 'The Old Schoolhouse' | 0..1 | [addressLine](#3406) |  |
| **Address line 2 (12823@0.0.1 )** | House number, dependent thoroughfare name and descriptor without commas, e.g. '23 Mill Lane' | 0..1 | [addressLine](#3406) |  |
| **Address line 3 (12824@0.0.1 )** | Dependent locality/village, e.g. 'Boxgrove' | 0..1 | [addressLine](#3406) |  |
| **Address line 4 (12825@0.0.1 )** | Post town, e.g. 'Leeds' | 0..1 | [addressLine](#3406) |  |
| **Address line 5 (12826@0.0.1 )** | County (if present), e.g. 'Hampshire', 'Hants' | 0..1 | [addressLine](#3406) |  |
| **Postcode (12827@0.0.1 )** | 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](#3412) |  |

#### Family History [1..1]

*One report containing Family History must be submitted together with each Registration report.*

This section should be collected for all participants to assist with interpretation of the genome data.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Mothers Ethnic Origin (12587)** | The ethnic origin of the participant's father, against supplied enumeration. | 1..1 | [ethnicCategory](#337) | Ethnicity (Genomics England Shared) |
| **Mothers Ethnic Origin Other (12588)** | Ethnic origin, if not in enumeration supplied | 0..1 | [xs:string](#38) |  |
| **Mothers Other Relevant Ancestry (12589)** | Pertinent to clinical findings, additional relevant ancestry such as ashkenazi | 0..1 | [xs:string](#38) |  |
| **Fathers Ethnic Origin (12595)** | The ethnic origin of the participant's father, against supplied enumeration. | 1..1 | [ethnicCategory](#337) | Ethnicity (Genomics England Shared) |
| **Fathers Ethnic Origin Other (12596)** | Father's ethnic origin, if not in enumeration supplied | 0..1 | [xs:string](#38) |  |
| **Fathers Other Relevant Ancestry (12597)** | Pertinent to clinical findings, additional relevant ancestry such as ashkenazi | 0..1 | [xs:string](#38) |  |

#### Clinical Information [1..1]

*One report containing Clinical Information must be submitted together with each Registration report.*

Please specify the participant type (proband versus family member). Based on the participant type either submit proband or relative datasets

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant Type (12554)** | Type of participant - rare disease proband or participating relative | 1..1 | [participantType](#33928) **Proband**:Proband **Relative**:Relative |  |

#### Information [1..1]

Record clinical information for either the Proband or Relative.

*One of the following must be submitted together with each Information report.*

##### Proband [1..1]

This section captures information on the proband’s family members, including affected individuals, parental consanguinity and pattern of recruitment (e.g. trio with mother and father).  
  
Please highlight in ‘Out of Area Recruitment’ whether any of the relatives are going to be recruited in a different GMC area from the proband.  
  
Indicate if incomplete penetrance (of a dominant trait) IN THIS FAMILY if known.  
  
Indicate if there are likely to be multiple monogenic causes of the phenotypes in the family. If these are cosegregating (including for example if they are all only present in the proband), select ‘Yes: cosegregating’. If they are segregating separately, select ‘Yes: segregating separately’.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | | **Data Type** | **Related To** |
| **Out of Area Recruitment (35540)** | Flag if a relative of the proband is recruited out of area | 1..1 | | [yesNo](#10099) **yes**:Yes **no**:No |  |
| **Participants in family (39048)** | The number of participants submitted as part of this family. If the family size changes before samples are submitted this information must be updated, as the family size is used to confirm that number of participants from whom samples are taken is correct prior to sequencing. | 1..1 | | [xs:positiveInteger](#53) |  |
| **Group Type (12555)** | The type of family group used against supplied enumeration | 1..1 | | [groupType](#33506) **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 |  |
| **Consanguinity (14564)** | This indicates that the participant is the product of a consanguinous relationship | 0..1 | | [consanguinity](#33290) **U**:Unknown **P**:Possible **N**:No **Y**:Yes |  |
| **Penetrance (31272)** | Is there any indication that the disease is not fully penetrant in this family? | 0..1 | | [penetrance](#33941) **yes**:Yes **no**:No **unknown**:Unknown |  |
| **Multiple Monogenic Causes (39051)** | Are there likely to be multiple monogenic causes of the phenotypes in this family? | 0..1 | | [MultipleMonogenicCauses](#39049) **Segregating**:Segregaing **Segregating\_separately**:Segregating Separately **No**:No |  |
| **Mother Affected (12558)** | Mother affected with same condition as proband, chosen from supplied enumeration | 1..1 | | [affectedStatus](#32990) **yes**:Yes **no**:No **unknown**:Unknown |  |
| **Father Affected (12559)** | Father affected with same condition as proband, chosen from supplied enumeration | 1..1 | | [affectedStatus](#32990) **yes**:Yes **no**:No **unknown**:Unknown |  |
| **Full Brothers Affected (12560)** | Number of full brothers with same condition | 1..1 | | [positiveInteger](#33993) |  |
| **Total Full Brothers (12561)** | Total number of full brothers | 1..1 | | [positiveInteger](#33993) |  |
| **Full Sisters Affected (12562)** | Number of full sisters with same condition | 1..1 | | [positiveInteger](#33993) |  |
| **Total Full Sisters (12563)** | Total number of full sisters | 1..1 | | [positiveInteger](#33993) |  |
| Rule | | | Participants in family and group type should be consistent i.e. if group is a trio then family size must be 3 | | |

###### Eligibility [1..1]

*One report containing Eligibility must be submitted together with each Proband report.*

Please state if the participant is suitable for recruitment according to the ‘Rare Diseases Eligibility Statements’, including details of the version used (e.g. version 1.4).

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Eligibility Statement Version (15017)** | Name and version of the disease specific eligibility statement - requested format [diseaseName]\_[eligibilityId] | 1..1 | [xs:string](#38) |  |
| **Eligible (15018)** | Participant's eligibility | 1..1 | [yesNo](#10099) **yes**:Yes **no**:No |  |

###### Diagnosis [1..\*]

*One or more reports containing Diagnosis must be submitted together with each Proband report.*

Please choose from the list of Genomics England rare diseases to indicate the participant’s phenotype. At least one disorder from this list should be included for every proband. Multiple diagnoses can be provided but each report should be submitted as its own event (i.e. contain its own event data and event reference, if available).  
  
For diagnoses that are not part of the Genomics England disorders, please use the ‘disease information (other)’ section using SNOMED CT/OMIM/ICD terms. This could be used in a number of situations, for example:  
• A recruited relative is unaffected with the proband’s disorder, but has another medical condition which it is important to include in their record.  
• A recruited proband or affected relative has a second disorder which isn’t on the list of Genomics England disorders, but may be relevant for interpretation of their genome.  
• A participant entered in a familial cancer category has more than one diagnosis of cancer which can be entered separately here.  
NOTE: HPO terms should be provided to supplement these diagnoses and/or where diagnosis cannot be captured within SNOMED CT/OMIM/ICD.

Event Details [1..1]

*One report containing Event Details must be submitted together with each Diagnosis report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

Disease Information [1..1]

Record disease information associated with the diagnosis. This will be provided as a choice element in the XML Schemas.

*One of the following must be submitted together with each Information report.*

Disease Information GEL [1..1]

Disease information classified according to Genomics England Rare Disease Conditions and Eligibility Criteria.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | | **Data Type** | **Related To** |
| **Disease Group (12580)** | Top-level classification of rare diseases (project specific). Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Disease Subgroup (12581)** | Narrower classification of disease. Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Specific Disease (12582)** | Specific rare disorder within this classification Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Age of Onset (12583)** | Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders. | 1..1 | | [ageInYearsFractions](#34395) |  |
| Rule | | | Disease group, subgroup Disease Group, Subgroup and Specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | | |

**And in the case of non-GEL disease information,**

Disease Information Other [1..1]

Diagnoses that are not part of the Genomics England disorders. This whole section is repeatable to allow submission of multiple diagnoses. For each diagnosis please submit the medical condition as either an ICD10, SnomedCT or OMIM Code together with the age of onset.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Age of Onset (12583)** | Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders. | 1..1 | [ageInYearsFractions](#34395) |  |

Medical Condition [1..1]

*One of the following must be submitted together with each Medical Condition report.*

Record the medical condition as either an ICD10 code, or a SNOMEDCT code or an OMIM code. This will be provided as a choice element in the XML Schemas.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Medical Condition ICD10 Code (31155)** | Medical condition coded using ICD10 | 1..1 | [ICD10Code](#33567) |  |
| **Or in the case of SnomedCT conditions,** | | | | |
| **Medical Condition SnomedCT Code (31153)** | Medical condition coded using SnomedCT | 1..1 | [diagnosticTermsSnomedCT](#33365) |  |
| **Or in the case of OMIM conditions,** | | | | |
| **OMIM Code (29827@0.0.1 )** | OMIM code that best describes disorder | 1..1 | [xs:string](#38) |  |

**Or in the case of a relative,**

##### Relative [1..1]

Select the relative’s biological relationship to the proband, choosing ‘other’ if not available in the drop-down list. To allow robust family linkage, please include full details of the proband’s name, date of birth, and NHS number.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Biological Relationship to Proband (12564)** | Biological Relationship to Proband | 1..1 | [biologicalFamilyRelationship](#33189) |  |
| **Other Biological Relationship to Proband (12565)** | Biological Relationship to Proband if not in the enumeration supplied | 0..1 | [xs:string](#38) |  |
| **Proband Forenames (33983)** | The forenames of the proband associated with the family group, used for consistency checks | 1..1 | [xs:string](#38) |  |
| **Proband Surname (33984)** | The surname of the proband associated with the family group, used for consistency checks | 1..1 | [xs:string](#38) |  |
| **Proband Date of Birth (33986)** | The date of birth of the proband associated with the family group, used for consistency checks | 1..1 | [xs:date](#58) |  |

Proband Person Identifier [1..1]

Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland) of the Proband. Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital. Used for consistency checks

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Proband NHS Number (33985)** | The nhs number of the proband associated with the family group, used for consistency checks | 1..1 | [xs:string](#38) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **Proband CHI Number (55382)** | The chi number of the proband associated with the family group, used for consistency checks | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Proband Health and Care Number (55383)** | The health and social care number of the proband associated with the family group, used for consistency checks | 1..1 | [healthAndCareNumber](#54236) |  |

###### Diagnosis [0..\*]

*Multiple reports containing Diagnosis can be submitted together with each Relative report.*

Please choose from the list of Genomics England rare diseases to indicate the participant’s phenotype. At least one disorder from this list should be included for every proband. Multiple diagnoses can be provided but each report should be submitted as its own event (i.e. contain its own event data and event reference, if available).  
  
For diagnoses that are not part of the Genomics England disorders, please use the ‘disease information (other)’ section using SNOMED CT/OMIM/ICD terms. This could be used in a number of situations, for example:  
• A recruited relative is unaffected with the proband’s disorder, but has another medical condition which it is important to include in their record.  
• A recruited proband or affected relative has a second disorder which isn’t on the list of Genomics England disorders, but may be relevant for interpretation of their genome.  
• A participant entered in a familial cancer category has more than one diagnosis of cancer which can be entered separately here.  
NOTE: HPO terms should be provided to supplement these diagnoses and/or where diagnosis cannot be captured within SNOMED CT/OMIM/ICD.

Event Details [1..1]

*One report containing Event Details must be submitted together with each Diagnosis report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

Disease Information [1..1]

Record disease information associated with the diagnosis. This will be provided as a choice element in the XML Schemas.

*One of the following must be submitted together with each Information report.*

Disease Information GEL [1..1]

Disease information classified according to Genomics England Rare Disease Conditions and Eligibility Criteria.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | | **Data Type** | **Related To** |
| **Disease Group (12580)** | Top-level classification of rare diseases (project specific). Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Disease Subgroup (12581)** | Narrower classification of disease. Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Specific Disease (12582)** | Specific rare disorder within this classification Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Age of Onset (12583)** | Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders. | 1..1 | | [ageInYearsFractions](#34395) |  |
| Rule | | | Disease group, subgroup Disease Group, Subgroup and Specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | | |

**Or in the case of non-GEL disease information,**

Disease Information Other [1..1]

Diagnoses that are not part of the Genomics England disorders. This whole section is repeatable to allow submission of multiple diagnoses. For each diagnosis please submit the medical condition as either an ICD10, SnomedCT or OMIM Code together with the age of onset.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Age of Onset (12583)** | Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders. | 1..1 | [ageInYearsFractions](#34395) |  |

Medical Condition [1..1]

*One report containing a medical condition must be submitted together with each Disease Information other report.*

Record the medical condition as either an ICD10 code, or a SNOMEDCT code or an OMIM code. This will be provided as a choice element in the XML Schemas.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Medical Condition ICD10 Code (31155)** | Medical condition coded using ICD10 | 1..1 | [ICD10Code](#33567) |  |
| **Or in the case of SnomedCT conditions,** | | | | |
| **Medical Condition SnomedCT Code (31153)** | Medical condition coded using SnomedCT | 1..1 | [diagnosticTermsSnomedCT](#33365) |  |
| **Or in the case of OMIM conditions,** | | | | |
| **OMIM Code (29827@0.0.1 )** | OMIM code that best describes disorder | 1..1 | [xs:string](#38) |  |

### Consent [1..1]

*One report containing Consent must be submitted together with each Registration and Consent report.*

This section reports information obtained at consent, including the overall consent status (consent given) and the individual questions and responses relating to the participant’s options regarding additional findings (Consent Details (29742.1)).  
  
Additional mandatory fields include the date of consent, full name of the person taking consent, and details of the version of the consent form and information sheet used for participation in the 100,000 Genomes Project.  
  
No further data should be entered if the answer to the ‘Consent Given’ question is ‘No’.

The Assent form can be used for children to sign to indicate their assent to join the project, but they must legally also have a parental consent form completed.   
  
If both members of a couple consent to receive additional findings together, their report will include an analysis of conditions where they are BOTH carriers of an autosomal recessive condition on the list of additional findings. This is recorded in the section below, where details of the other partner are required to generate the right couple report.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Name and Version of Consent Form (34549)** | Name and Version of form used. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 1..1 | [xs:string](#38) |  |
| **Consent Given (12545)** | Yes no answer to consent given | 1..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |
| **Consent Form (12546)** | File name of uploaded PDF copy of consent form - requested format [ParticipantId]\_consent\_[TimeStamp].pdf | 0..1 | [xs:string](#38) |  |
| **Person Taking Consent (12547)** | The full name of the person taking consent | 1..1 | [xs:string](#38) |  |
| **Name and Version of Participant Information Sheet (4454)** | Name and Version of information sheet presented. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 1..1 | [xs:string](#38) |  |
| **Name and Version of Assent Form (34552)** | Name and Version of Cancer Assent form used. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 0..1 | [xs:string](#38) |  |
| **Assent Form (34543)** | File name of the uploaded PDF copy of the assent form. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 0..1 | [xs:string](#38) |  |
| **Additional optional consent materials (40373)** | Names and versions of consent additional consent materials used. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 0..\* | [xs:string](#38) |  |

#### Event Details [1..1]

*One report containing Event Details must be submitted together with each Consent report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

#### Consent Details [1..1]

*One report containing Consent Details must be submitted together with each Consent report.*

Details corresponding to the questions and responses on the consent form.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Health Related Additional Findings (34544)** | Health-related additional findings: Does the participant want these looked for and fed back to their clinical team? | 0..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |
| **Reproductive Additional Findings (34546)** | Reproductive additional findings: Does the participant want these looked for and fed back to their clinical team? | 0..1 | [yesNoNotRelevant](#54322) **yes**:yes **no**:no **not\_relevant**:not relevant |  |
| **Couple Report Requested (39077)** | If a couple are joining the project together and are potentially planning further children in the future, have they both consented to receive additional reproductive findings? | 0..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |

##### Couple Report Details [0..1]

*One report containing Couple Report Details can be submitted together with each Consent report.*

If a couple report is requested (yes), then complete the following:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Partner Participant ID (39078)** | The Genomics England participant ID of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [participantId](#54269) |  |
| **Partner Forename (39080)** | The forenames of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [xs:string](#38) |  |
| **Partner Surname (39081)** | The surname of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [xs:string](#38) |  |
| **Partner Date of Birth (39083)** | The date of birth of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [xs:date](#58) |  |

###### Partner Person Identifier [1..1]

*One report containing Partner Person Identifier Report Details must be submitted together with each Couple Report Details report.*

Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Partner NHS Number (39082)** | The nhs number of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [nhsNumber](#300) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **Partner CHI Number (55378)** | The chi number of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Partner Health and Care Number (55379)** | The health and care number of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [healthAndCareNumber](#54236) |  |

# Core Data

Data items in this section represent core clinical data relevant to the individual rare disease participant. Every participant who is affected with a disorder on the Genomics England rare disease list must have:  
• Phenotype data collected using the ‘data model’ (list of HPO terms) specific to their disorder.  
• Clinical test data entered for all of the clinical tests listed as relevant for their disorder (see separate spreadsheet).  
• Genetic test results entered for all prior genetic testing (whether positive, negative or of uncertain significance)  
• A participant who has an additional disorder not on the Genomics England rare disease list can have clinical data entered if this is likely to be relevant to the interpretation of their genome results; this is optional:  
• Phenotype data can be collected using HPO terms in the ‘free HPO’ section of one of the specific data models. Ideally a data model should be chosen which is similar to the patient’s disorder, and a discrepancy flag should be used to indicate that this is not the actual diagnosis.  
• Clinical test data can be entered for any participant from the available list of clinical tests, including positive and negative results.  
• Genetic test results can be entered for any participant, including positive and negative results.  
• Pedigree diagram is required for each proband  
Relatives who are unaffected with any disorder can still have relevant clinical test or genetic test results uploaded, particularly if there are relevant negative results (eg normal echocardiogram in the parent of a child with congenital heart disease); this is optional.  
All reports of core clinical data must include the following shared data elements.

## Pedigree Data

TBC

## Consent Update

This section reports changes in a participant’s consent status after they have joined the project, including changes in preference relating to the participant’s options regarding additional findings (Consent Details (29742.1)).  
  
If a patient withdraws from the project, please complete a Withdrawal form, not a Consent update form.

The Assent form can be used for children to sign to indicate their assent to join the project, but they must legally also have a parental consent form completed.

If both members of a couple consent to receive additional findings together, their report will include an analysis of conditions where they are BOTH carriers of an autosomal recessive condition on the list of additional findings. This is recorded in the section below, where details of the other partner are required to generate the right couple report.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Name and Version of Consent Form Update (34558)** | Name and Version of cancer update form used. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 1..1 | [xs:string](#38) |  |
| **Consent Given (12545)** | Yes no answer to consent given | 1..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |
| **Consent Form (12546)** | File name of uploaded PDF copy of consent form - requested format [ParticipantId]\_consent\_[TimeStamp].pdf | 0..1 | [xs:string](#38) |  |
| **Person Taking Consent (12547)** | The full name of the person taking consent | 1..1 | [xs:string](#38) |  |
| **Name and Version of Participant Information Sheet Update (34564)** | Name and Version of information sheet presented. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 1..1 | [xs:string](#38) |  |
| **Name and Version of Assent Form (34552)** | Name and Version of Cancer Assent form used. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 0..1 | [xs:string](#38) |  |
| **Assent Form (34543)** | File name of the uploaded PDF copy of the assent form. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 0..1 | [xs:string](#38) |  |
| **Additional optional consent materials (40373)** | Names and versions of consent additional consent materials used. Please see appendix F for latest list of consent forms, participant information sheets, additional optional consent materials and enumerations. | 0..1 | [xs:string](#38) |  |

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Consent Update report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### Event Details [1..1]

*One report containing Event Details must be submitted together with each Consent Update report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

### Consent Details [1..1]

*One report containing Consent Details must be submitted together with each Consent Update report.*

Details corresponding to the questions and responses on the consent form.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Health Related Additional Findings (34544)** | Health-related additional findings: Does the participant want these looked for and fed back to their clinical team? | 0..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |
| **Reproductive Additional Findings (34546)** | Reproductive additional findings: Does the participant want these looked for and fed back to their clinical team? | 0..1 | [yesNoNotRelevant](#54322) **yes**:yes **no**:no **not\_relevant**:not relevant |  |
| **Couple Report Requested (39077)** | If a couple are joining the project together and are potentially planning further children in the future, have they both consented to receive additional reproductive findings? | 0..1 | [yesNo](#54321) **yes**:Yes **no**:No |  |

#### Couple Report Details [0..1]

*One report containing Couple Report Details can be submitted together with each Consent Details report.*

If a couple report is requested (yes), then complete the following:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Partner Participant ID (39078)** | The Genomics England participant ID of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [participantId](#54269) |  |
| **Partner Forename (39080)** | The forenames of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [xs:string](#38) |  |
| **Partner Surname (39081)** | The surname of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [xs:string](#38) |  |
| **Partner Date of Birth (39083)** | The date of birth of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [xs:date](#58) |  |

##### Partner Person Identifier [1..1]

*One report containing Partner Person Identifier Report Details must be submitted together with each Couple Report Details report.*

Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Partner NHS Number (39082)** | The nhs number of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [nhsNumber](#300) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **Partner CHI Number (55378)** | The chi number of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Partner Health and Care Number (55379)** | The health and care number of the partner with whom this participant is to receive a couple report, used to report autosomal recessive carrier status | 1..1 | [healthAndCareNumber](#54236) |  |

## Phenotype Report

Please provide information about who entered the data and when, to ensure traceability of the data source.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Clinical Assessment Letter (30853)** | File name of uploaded copy of the most informative letter(s) or assessment relating to the patients phenotype - requested format [ParticipantId]\_assessment\_[TimeStamp] | 0..\* | [xs:string](#38) |  |
| **Phenotype Report Code (33567)** | 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](#38) |  |

### Event Details [1..1]

*One report containing Event Details must be submitted together with each Phenotype report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

### Consultant Details [0..1]

*One report containing Consultant Details can be submitted together with each Phenotype report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Full Name of Responsible Consultant (12774)** | Nominated person responsible for patients clinical care and recipient of clinical reports and communications for Genomics England | 1..1 | [xs:string](#38) |  |
| **Consultant GMC number (31254)** | GMC number of consultant with responsibility for the patient's clinical care | 1..1 | [consultantCode](#32403) | CONSULTANT CODE (TREATMENT) (CR0660 from Cancer Outcomes and Services Dataset) |
| **Full Name not Consultant (4495)** | Full name of person entering data on behalf of consultant | 0..1 | [xs:string](#38) |  |
| **Contact number (14520)** | Phone number for the consultant. | 0..1 | [ukTelephoneNumber](#54308) |  |
| **Hospital of Responsible Consultant (12516)** | ODS code of the hospital to which the consultant is contracted under their MAIN SPECIALTY for the purposes of the current work. | 0..1 | [organisationSiteCode](#42015) |  |

### Phenotype Statement [1..\*]

*One or more reports containing Phenotype Statements must be submitted for each Phenotype report.*

Phenotype statements (coded in the Human Phenotype Ontology or HPO) are grouped into ‘data models’ for each rare disorder on the Genomics England list.  
  
The HPO is a structured hierarchy that can be used to describe medical signs or symptoms. An example of how this works is shown in Figure 2. In this case, if you know that the patient has ‘multiple medullary renal cysts’, this gives you a very specific piece of information about their kidney problem, which provides a high level of information content to use when analysing their genome sequence. If you knew that the patient had renal cysts, but you didn’t have the biopsy result to show exactly what type, you could choose a higher term such as ‘renal cyst’ which contains slightly less information, but still captures an important part of the patient’s condition.

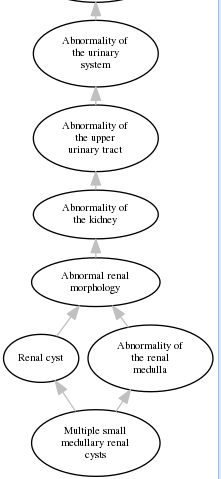


Figure 2: Example of the hierarchical relationship between terms found in the HPO  
Knowing that a patient DOESN’T have ‘multiple small medullary renal cysts’ is much less informative, although there might be certain conditions where it is important to know that this specific feature is absent. In general however, it would be more useful to know that the patient doesn’t have any ‘abnormality of the kidney’, or even that they don’t have ‘renal cyst’, as this gives a much larger amount of information about their clinical condition.  
It is therefore helpful to collect POSITIVE information about the phenotype at as SPECIFIC a level as possible, while collecting NEGATIVE (or absent) information at as GENERAL a level as possible. It is important not to collect information at too high a level though, because the higher terms in the ontology have a lot of terms below them (each of the higher terms shown in Figure 2 is related in the tree to many others which aren’t shown in this limited diagram), so stating that a patient doesn’t have an ‘abnormality of the urinary system’, for example, would mean that every term in that section of the ontology would be considered as negative in the analysis, and you might not have enough information available about the patient to know that this is true. It might also conflict with another phenotype term that you have stated as present, because phenotype terms can have multiple higher-level parent terms in different body systems  
  
Modifiers in HPO  
The HPO ontology contains a standard set of modifying terms: laterality, onset, progression, severity and spatial pattern. Each modifier relates to a single HPO term in that individual. Not all modifiers will be relevant for each HPO term, but multiple modifiers can be applied to each HPO term where this adds to the phenotypic description.  
  
Laterality  
The options given are bilateral, left, right or unilateral.  
  
Onset  
This refers to the age of onset of the feature described by that particular HPO term, not the overall clinical presentation in the patient. The definition for the ‘onset’ categories is given in Table 1 below.  
  
  
Age of onset modifier  
HPO code  
Definition  
Congenital onset\*  
  
HP:0003577  
A phenotypic abnormality that is present at birth  
  
Embryonal onset\*  
HP:0011460  
Onset of disease at up to 8 weeks of gestation  
  
Fetal onset\*  
HP:0011461  
Onset prior to birth but after 8 weeks of gestation  
Neonatal onset  
  
HP:0003623  
Onset of signs or symptoms of disease within the first 28 days of life  
Infantile onset  
  
HP:0003593  
Onset of signs or symptoms of disease between 28 days to one year of life  
Childhood onset  
  
HP:0011463  
Onset of disease at the age of between 1 and 5 years.  
Juvenile onset  
  
HP:0003621  
Onset of signs or symptoms of disease between the age of 5 and 15 years  
Adult onset\*\*  
  
HP:0003581  
Onset of disease manifestations in adulthood, defined here as at the age of 16 years or later  
  
Young adult onset\*\*  
HP:0011462  
Onset of disease at the age of between 16 and 40 years  
  
Middle age onset\*\*  
HP:0003596  
A type of adult onset with onset of symptoms at the age of 40 to 60 years  
  
Late onset\*\*  
HP:0003584  
A type of adult onset with onset of symptoms after the age of 60 years  
Table 1: HPO Age of Onset definitions  
\* Embryonal onset and Fetal onset are subgroups of Congenital onset  
\*\* Young adult, Middle age and Late onset are all subgroups of Adult onset  
  
Progression  
The options are non-progressive or progressive.  
  
Severity   
The options are borderline, mild, moderate, severe or profound.  
  
Spatial pattern  
The options are distal, generalised, localised or proximal.  
  
Free entry of HPO terms  
In addition to the suggested HPO terms in the disorder-specific data model, it is also possible to add HPO terms using a search function which can access the whole ontology. These terms should be used to add rare or additional features of the phenotype; this can include features which are not thought to be associated with the presenting condition. Occasionally this will be the primary means of data collection, for example if the patient has a very unusual condition recruited under the ‘Ultra-rare undescribed conditions’ category.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **HPO Build Number (14643)** | This is the build number of the HPO ontology used. | 0..1 | [xs:string](#38) |  |
| **Phenotype Identifier (14642)** | The identity of the statement within the Human Phenotype Ontology | 1..1 | [hpoID](#33564) |  |
| **Phenotype Present (14646)** | Presence of the phenotypic abnormality. | 1..1 | [Present](#34008) **unknown**:Unknown **yes**:Yes **no**:No |  |

#### Modifier [0..\*]

*A maximum of one report containing Modifiers can be submitted together with each Phenotype Statement report. The Phenotype Statement report can be submitted without this information.*

If applicable, modifiers can be recorded against each phenotype statement. These should be recorded using the subclasses of modifiers in HPO e.g. if a position was being recorded, the modifier id would be "HP:0012833" and the modifier would be "Unilateral". More than one modifier can be recorded for each phenotype statement.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Modifier Identifier (55368)** | HPO Modifier Identifier. This should be consistent with the HPO build number. Only valid HPO codes will be accepted. | 1..1 | [hpoID](#33564) |  |
| **Modifier (55369)** | HPO Modifier Name. This should be consistent with the HPO build number and the HPO Modifier ID. Only valid HPO modifiers will be accepted. | 1..1 | [xs:string](#38) |  |

## Withdrawals

This section should be used to record patient withdrawal from the project which occurs after the time of initial data collection.

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Withdrawals report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### Withdrawal [1..\*]

*One or more reports containing Withdrawal must be submitted for each Withdrawals report.*

This section should be used to record patient withdrawal from the project which occurs after the time of initial data collection.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Withdrawal Form (12730)** | Filename of uploaded copy of scanned withdrawal form pdf - requested format is [ParticipantId]\_withdrawal\_[TimeStamp].pdf | 0..1 | [xs:string](#38) |  |
| **Withdrawal Option (12728)** | Indicating full or partial withdrawal | 1..1 | [consentWithdrawalOptions](#54192) **full\_withdrawal**:OPTION 2: FULL WITHDRAWAL: No further use **partial\_withdrawal**:OPTION 1: PARTIAL WITHDRAWAL: No further contact |  |
| **Name and Version of the Withdrawal Form Used (12729)** | Name and Version of form used - list of names and versions available from genomicsengland.co.uk/library-and-resources/ | 1..1 | [genomicsEnglandConsentWithdrawalForms](#54226) **6a**:Withdrawal information and form – for adult or child participants (6a) **6b**:Consultee declaration of advice regarding adult participant withdrawal information – for consultees (withdrawal) (6b) |  |
| **Person Reporting Withdrawal (12731)** | Full name, including forenames and surname, of person reporting withdrawal. | 1..1 | [xs:string](#38) |  |

#### Event Details [1..1]

*One report containing Event Details must be submitted together with each Withdrawal report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

## Diagnoses

Please choose from the list of Genomics England rare diseases to indicate the participant’s phenotype. At least one disorder from this list 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 (if available). For diagnoses that are not part of the Genomics England disorders, please use the ‘disease information (other)’ section using SNOMED CT/OMIM/ICD terms. This could be used in a number of situations, for example: • A recruited relative is unaffected with the proband’s disorder, but has another medical condition which it is important to include in their record. • A recruited proband or affected relative has a second disorder which isn’t on the list of Genomics England disorders, but may be relevant for interpretation of their genome. • A participant entered in a familial cancer category has more than one diagnosis of cancer which can be entered separately here. NOTE: HPO terms should be provided to supplement these diagnoses and/or where diagnosis cannot be captured within SNOMED CT/OMIM/ICD.

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Diagnoses report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### Diagnosis [1..\*]

*One or more reports containing Diagnosis must be submitted together with each Diagnoses report.*

Please choose from the list of Genomics England rare diseases to indicate the participant’s phenotype. At least one disorder from this list should be included for every proband. Multiple diagnoses can be provided but each report should be submitted as its own event (i.e. contain its own event data and event reference, if available).  
  
For diagnoses that are not part of the Genomics England disorders, please use the ‘disease information (other)’ section using SNOMED CT/OMIM/ICD terms. This could be used in a number of situations, for example:  
• A recruited relative is unaffected with the proband’s disorder, but has another medical condition which it is important to include in their record.  
• A recruited proband or affected relative has a second disorder which isn’t on the list of Genomics England disorders, but may be relevant for interpretation of their genome.  
• A participant entered in a familial cancer category has more than one diagnosis of cancer which can be entered separately here.  
NOTE: HPO terms should be provided to supplement these diagnoses and/or where diagnosis cannot be captured within SNOMED CT/OMIM/ICD.

#### Event Details [1..1]

*One report containing Event Details must be submitted together with each Diagnosis report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

#### Disease Information [1..1]

Record disease information associated with the diagnosis. This will be provided as a choice element in the XML Schemas.

*One of the following must be submitted together with each Information report.*

##### Disease Information GEL [1..1]

Disease information classified according to Genomics England Rare Disease Conditions and Eligibility Criteria.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | | **Data Type** | **Related To** |
| **Disease Group (12580)** | Top-level classification of rare diseases (project specific). Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Disease Subgroup (12581)** | Narrower classification of disease. Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Specific Disease (12582)** | Specific rare disorder within this classification Disease group, subgroup and specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | 1..1 | | [xs:string](#38) |  |
| **Age of Onset (12583)** | Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders. | 1..1 | | [ageInYearsFractions](#34395) |  |
| Rule | | | Disease group, subgroup Disease Group, Subgroup and Specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List. | | | |

**Or in the case of non-GEL disease information,**

##### Disease Information Other [1..1]

Diagnoses that are not part of the Genomics England disorders. This whole section is repeatable to allow submission of multiple diagnoses. For each diagnosis please submit the medical condition as either an ICD10, SnomedCT or OMIM Code together with the age of onset.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Age of Onset (12583)** | Age of onset of predominant features in years (fractions). Please use negative numbers for prenatal disorders. | 1..1 | [ageInYearsFractions](#34395) |  |

###### Medical Condition [1..1]

*One report containing a medical condition must be submitted together with each Disease Information Other report.*

Record the medical condition as either an ICD10 code, or a SNOMEDCT code or an OMIM code. This will be provided as a choice element in the XML Schemas.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Medical Condition ICD10 Code (31155)** | Medical condition coded using ICD10 | 1..1 | [ICD10Code](#33567) |  |
| **Or in the case of SnomedCT conditions,** | | | | |
| **Medical Condition SnomedCT Code (31153)** | Medical condition coded using SnomedCT | 1..1 | [diagnosticTermsSnomedCT](#33365) |  |
| **Or in the case of OMIM conditions,** | | | | |
| **OMIM Code (29827@0.0.1 )** | OMIM code that best describes disorder | 1..1 | [xs:string](#38) |  |

## Investigations

Investigation reports can include observations, findings and laboratory tests.

Observations, findings and laboratory tests can be submitted as attributes of investigation reports using the models included below.

Please refer to the Rare Disease Investigations catalogue where requirements for Genomics England specific reports are specified.

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Investigations report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### General Investigation Report [0..\*]

*One or more reports containing Investigation Reports must be submitted together with each Investigations report.*

Investigation reports can include multiple findings, clinical tests and qualifiers.

#### Event Details [1..1]

*One report containing Event Details must be submitted together with each Investigations report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

#### Finding [0..\*]

*Multiple reports containing findings can be submitted together with each Investigations report.*

Record clinical finding, observation or result, if applicable.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Start Date Time (53791)** | Date and time the event started | 0..1 | [xs:dateTime](#59) |  |
| **Duration (53792)** | Duration of clinical event i.e. duration of observation, lab test, procedure or therapy | 0..1 | [xs:duration](#61) |  |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to e.g. SNOMED CT | 0..1 | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to e.g. 21.0.0 RF2 | 0..1 | [xs:string](#38) |  |
| **Finding Type ID (53795)** | Identifier or Code within the chosen data standard for the finding | 0..1 | [xs:string](#38) |  |
| **Finding Type (53796)** | Name of the type of finding or type or laboratory test | 1..1 | [xs:string](#38) |  |
| **Unit of Measurement (53797)** | Unit of measurement for finding recorded as UCUM codes | 0..1 | [xs:string](#38) |  |
| **Range (53798)** | Upper and Lower range of results for the finding | 0..1 | [xs:string](#38) |  |
| **Comments (53799)** | Any additional comments related to the finding | 0..1 | [xs:string](#38) |  |
| **Finding (53800)** | Clinical finding or result of laboratory test | 1..1 | [xs:string](#38) |  |
| **Patient Status (34004@0.0.1 )** | Represents the patient's status when test was performed | 0..1 | [patientStatus](#33934) **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 |  |
| **Assessment (29524@0.0.1 )** | Assessment of findings and clinical significance | 0..1 | [clinicalTestAbnormality](#33277) **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@0.0.1 )** | File name of uploaded copy of clinical report - requested format [Participant ID]\_[Local Report Identifier] | 0..1 | [xs:string](#38) |  |

##### Qualifier [0..\*]

*Multiple reports containing Qualifiers can be submitted together with each Findings report.*

Qualifier for clinical finding or report. e.g. laterality.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to e.g. SNOMED CT | 0..unbounded | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to e.g. 21.0.0 RF2 | 0..unbounded | [xs:string](#38) |  |
| **Qualifier Type ID (55340)** | Identifier for the qualifier type | 1..1 | [xs:string](#38) |  |
| **Qualifier Type (55338)** | Name of the qualifier | 1..1 | [xs:string](#38) |  |
| **Qualifier (55339)** | Value of the qualifier | 1..1 | [xs:string](#38) |  |

#### Laboratory Test [0..\*]

*Multiple reports containing Laboratory tests can be submitted together with each Investigations report.*

Record laboratory test result, if applicable.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Start Date Time (53791)** | Date and time the event started | 0..1 | [xs:dateTime](#59) |  |
| **Duration (53792)** | Duration of clinical event i.e. duration of observation, lab test, procedure or therapy | 0..1 | [xs:duration](#61) |  |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to e.g. SNOMED CT | 0..1 | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to e.g. 21.0.0 RF2 | 0..1 | [xs:string](#38) |  |
| **Finding Type ID (53795)** | Identifier or Code within the chosen data standard for the finding | 0..1 | [xs:string](#38) |  |
| **Finding Type (53796)** | Name of the type of finding or type or laboratory test | 1..1 | [xs:string](#38) |  |
| **Unit of Measurement (53797)** | Unit of measurement for finding recorded as UCUM codes | 0..1 | [xs:string](#38) |  |
| **Range (53798)** | Upper and Lower range of results for the finding | 0..1 | [xs:string](#38) |  |
| **Comments (53799)** | Any additional comments related to the finding | 0..1 | [xs:string](#38) |  |
| **Finding (53800)** | Clinical finding or result of laboratory test | 1..1 | [xs:string](#38) |  |
| **Patient Status (34004@0.0.1 )** | Represents the patient's status when test was performed | 0..1 | [patientStatus](#33934) **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@0.0.1 )** | File name of uploaded copy of clinical report - requested format [Participant ID]\_[Local Report Identifier] | 0..1 | [xs:string](#38) |  |
| **Assessment (29524@0.0.1 )** | Assessment of findings and clinical significance | 0..1 | [clinicalTestAbnormality](#33277) **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 |  |
| **Time Aspect (29880@0.0.1 )** | Indicate any time aspect of specimen collection, e.g. spot or 24 hour sampling | 0..1 | [timeAspect](#34220) **Spot**:Spot **Unspecified**:Unspecified **24hr Collection**:24hr Collection **Basal**:Basal - Time Course **Stimulated Peak**:Stimulated Peak - Time Course |  |
| **Sample Preconditions (53805)** | Indicate any preconditions applicable to sampling, for example fasting status | 0..1 | [samplingPreconditions](#34111) **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 |  |
| **Specimen (53806)** | Specimen type | 0..1 | [xs:string](#38) |  |
| **DateTime Specimen Signed Out (53811)** | Date and time specimen signed out | 0..1 | [xs:dateTime](#59) |  |
| **DateTime Specimen Collected (53812)** | Date and time specimen was collected | 0..1 | [xs:dateTime](#59) |  |
| **Test Status (39068@0.0.1 )** | Event status of the test or finding | 0..1 | [testStatus](#39067) **not\_done**:Not done **failed**:Failed **complete**:Complete **pending**:Pending |  |
| **Culture Site (53807)** | Microbiology Culture Site measurement. Recorded 1:1 | 0..1 | [xs:string](#38) |  |
| **Organism (53808)** | Microbiology Culture organism measurement. Recorded 1:1 | 0..1 | [xs:string](#38) |  |
| **Sensitivity (55365)** | Microbiology Culture sensitivity measurement. Recorded 1:1 | 0..1 | [xs:string](#38) |  |

##### Qualifier [0..\*]

*Multiple reports containing Qualifiers can be submitted together with each Laboratory Test report.*

Qualifier for clinical finding or report. e.g. laterality.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to e.g. SNOMED CT | 0..unbounded | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to e.g. 21.0.0 RF2 | 0..unbounded | [xs:string](#38) |  |
| **Qualifier Type ID (55340)** | Identifier for the qualifier type | 1..1 | [xs:string](#38) |  |
| **Qualifier Type (55338)** | Name of the qualifier | 1..1 | [xs:string](#38) |  |
| **Qualifier (55339)** | Value of the qualifier | 1..1 | [xs:string](#38) |  |

### Genetic Results [0..\*]

*Multiple reports containing Genetic Results can be submitted together with each Investigations report.*

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

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Sample Tissue of Origin (33274@0.0.1 )** | Origin of the sample tissue | 1..1 | [tissueOrigin](#34224) **Blood**:Blood **Skin**:Skin **Saliva**:Saliva **Muscle**:Muscle **CVS**:CVS **Amniocentesis**:Amniocentesis **Tumour**:Tumour **Other**:Other |  |
| **Sample Tissue of Origin Details (39065@0.0.1 )** | If sample tissue of origin is ‘other’, please provide details | 0..1 | [xs:string](#38) |  |
| **Assessment (29524@0.0.1 )** | Assessment of findings and clinical significance | 0..1 | [clinicalTestAbnormality](#33277) **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 |  |

#### Genetic Result [1..200]

*One or more reports containing Genetic Results can be submitted together with each Genetic Results report.*

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.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Genetic Test Laboratory (4563)** | Was this test performed in a diagnostic or research laboratory? | 0..1 | [geneticTestLaboratory](#54220) **research\_laboratory**:Research laboratory **diagnostic\_laboratory**:Diagnostic laboratory |  |
| **Test Scope (6101)** | The gene coded according to HGNC. Enter ‘genomewide’ if genomewide, e.g. karyotype or aCGH. | 1..1 | [geneScope](#54216) |  |
| **Scope Qualifiers (12764)** | 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](#38) |  |
| **Method of Test (12765)** | 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](#54221) |  |
| **Test Result (12744)** | (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 | [molecularTestResult](#54250) **normal**:Normal (negative) **fail**:Fail **abnormalitydetected**:Pathogenic abnormality detected **vus**:Variant of unknown significance detected |  |
| **Abnormal Molecular Result (14900)** | Record the details of the abnormal genotype using Genomic Coordinates | 0..1 | [xs:string](#38) |  |
| **Genome Build (34224)** | Record the relevant human genome build if an abnormal genotype is specified if applicable | 0..1 | [xs:string](#38) |  |
| **Abnormal Cytogenetic Result (34225)** | Record the details of the cytogenetic abnormality using IGCN standards | 0..1 | [xs:string](#38) |  |

#### Event Details [1..1]

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

#### Sample Details [0..1]

All sample investigations relating to germline or tumour molecular genetics should have the following:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Local Sample ID (12762)** | The local identifier for the source sample | 0..1 | [xs:string](#38) |  |
| **Sample Taken Date (12760)** | The date upon which the sample was taken | 0..1 | [xs:date](#58) | SAMPLE COLLECTION DATE (CR1010 from Cancer Outcomes and Services Dataset) |
| **Sample Receipt Date (12761)** | The date upon which the sample was received at the laboratory | 0..1 | [xs:date](#58) | SAMPLE RECEIPT DATE (CR0770 from Cancer Outcomes and Services Dataset) |

#### Genetic Reports [1..1]

If a genetic report was produced, then the initial list of five items should be extended with the following items.  
GUIDANCE: Upload all genetic test reports from this sample.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Description of Genetic Test (15153@0.0.1 )** | 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](#38) |  |
| **Genetic Report (15027@0.0.1 )** | File name of uploaded copy of report - requested format  [ParticipantId]\_molecular\_report\_[TimeStamp] | 1..unbounded | [xs:string](#38) |  |

**The following reports and relevant attributes are described in further detail in the Genomics England Investigations catalogue.**

* General Observations
  + Growth Parameters
  + Age of onset under 16
    - Pregnancy details
    - Birth details
    - Developmental milestones
* Patient History
  + Age at transplant loss
  + Age at diagnosis of chronic kidney disease
  + Age at ESRD
  + Inflammation metrics
  + Age of onset of motor symptoms
  + Infection metrics
  + APGAR score
  + Lifestyle factors
  + Renal history
* Exercise test
  + Childhood Health Assessment
  + Childhood Myositis Assessment
  + Intelligence Quotient Assessment
  + Performance Intelligence Quotient assessment
  + Ataxia assessment
  + Spastic Paraplegia Assessment
  + Manual Muscle Testing 8 (MMT8) Assessment
  + VAS Assessment
  + Development Quotient Assessment
  + Beighton test
* Vital signs
  + Blood pressure
  + Additional body measurements
  + Weight
* Biopsy
  + General Biopsy
  + Renal Biopsy
  + Nasal Cilia Imaging
* Imaging Diagnostics
  + General Imaging Diagnostics
  + Echocardiogram
  + Kidney Imaging
  + Liver Imaging
  + Facial features most in keeping with an OMIM disease
  + Heart/liver Iron measurement
  + Cardiac MRI
  + Doppler Diastolic function assessment
* Laboratory Test Report
  + General Laboratory Test Report
  + Urine Test
  + Urine Dip
  + Culture
  + Autoantibodies
  + TORCH screen
  + Arterial blood gas
  + Bone profile
  + Full Blood Count
  + Liver biochemistry
  + Pancreatic autoantibodies
  + Serum immunoglobulins
  + CSF tests
  + Renal biochemistry
  + Blood Tests
    - Biotinidase
    - Clotting
    - Coeliac antibodies
    - Complement
    - Congenital Myaesthenia Antibodies
    - Cortisol
    - Extended haematology investigations
    - Extended renal biochemistry
    - Glucose
    - Growth hormones
    - Hormones (other)
    - Inflammatory markers
    - Insulin and C-peptide
    - Lipids
    - Metabolic biochemistry
    - Microbiology antibodies
    - Porphyria investigations
    - Primary immunodeficiency investigations
    - Renin and aldosterone
    - Thyroid function testing
    - Virology
    - Vitamin B12
    - Sex hormones
  + Other enzymes
  + Fecal test
  + Venous blood gas
* Non-imaging Diagnostics
  + General Non-imaging Diagnostics
  + Heart observations
  + Forced vital capacity
  + ECG diagnostics
  + Sleep test
  + Cardiac Drug Challenge
  + Holter monitor test
  + Signal averaged ECG
  + Exercise test - cardiac
  + Electrophysiological study
  + Auditory Brainstem Response
  + Otoacoustic Emissions
  + Ocular metrics
  + Ocular Malformation Metrics
  + Visual Field
  + Ocular Pressure
  + Colour Plate Test
  + Electro-oculogram
  + Dark Adaptation Test
  + Electroretinogram
  + Visual Acuity
  + Refraction Error

## 

## Interventions

Intervention reports can include procedures and therapies / regimes.

Therapies, regimes and procedures can be submitted as attributes of intervention reports using the models included below.

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Interventions report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### Intervention Report [1..\*]

*One or more reports containing Interventions must be submitted together with each Interventions report.*

Intervention reports can include multiple event procedures and/or therapy/regimes.

#### Event Details [1..1]

*One report containing Event Details must be submitted together with each Interventions Report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

#### Procedure [0..\*]

Information regarding procedure

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to e.g. SNOMED CT | 1..1 | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to e.g. 21.0.0 RF2 | 1..1 | [xs:string](#38) |  |
| **Procedure ID (55342)** | Identifier for the procedure | 1..1 | [xs:string](#38) |  |
| **Procedure (55345)** | Name of the procedure | 1..1 | [xs:string](#38) |  |
| **Main Specialty Code (29100)** | The main specialty code of the consultant performing the intervention. | 0..1 | [specialty](#51989) |  |
| **Comments (5182)** | Follow-up comments | 0..1 | [xs:string](#38) |  |
| **Primary (55346)** | Flag to indicate if the procedure was a primary procedure. | 0..1 | [xs:boolean](#34) **0**:False **1**:True **true**:True **false**:False |  |
| **Start Date Time (53791)** | Date and time the event started | 1..1 | [xs:dateTime](#59) |  |
| **Duration (53792)** | Duration of clinical event i.e. duration of observation, lab test, procedure or therapy | 0..1 | [xs:duration](#61) |  |

##### Qualifier [0..\*]

*Multiple reports containing Qualifiers can be submitted together with each Procedure report.*

Qualifier for clinical finding or report. i.e. laterality.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to i.e. SNOMED CT | 0..unbounded | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to i.e. 21.0.0 RF2 | 0..unbounded | [xs:string](#38) |  |
| **Qualifier Type ID (55340)** | Identifier for the qualifier type | 1..1 | [xs:string](#38) |  |
| **Qualifier Type (55338)** | Name of the qualifier | 1..1 | [xs:string](#38) |  |
| **Qualifier (55339)** | Qualifier | 1..1 | [xs:string](#38) |  |

#### Therapy Regime [0..\*]

*Multiple reports containing Therapy Regimes can be submitted together with each Therapy Regime report.*

Information about therapies or regimes

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to e.g. SNOMED CT | 1..1 | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to e.g. 21.0.0 RF2 | 1..1 | [xs:string](#38) |  |
| **Therapy ID (12799)** | ID or Code within the chosen data standard for the therapy | 1..1 | [xs:string](#38) |  |
| **Therapy Name (29855)** |  | 0..1 | [xs:string](#38) |  |
| **Dose (12800)** | Dose prescribed according to the dictionary of medications and devices (http://www.nhsbsa.nhs.uk/1121.aspx) | 0..1 | [xs:string](#38) |  |
| **Frequency (12801)** | Frequency prescribed | 0..1 | [xs:string](#38) |  |
| **Intent (55347)** | Intent of the proposed treatment. | 0..1 | [treatmentIntent](#55348) **C**:Curative **Z**:Non Curative **X**:No active treatment **9**:Not Known |  |

##### Qualifier [0..\*]

*Multiple reports containing Qualifiers can be submitted together with each Procedure report.*

Qualifier for clinical finding or report. i.e. laterality.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Data Standard (53793)** | The data standard that the finding and coding corresponds to i.e. SNOMED CT | 0..unbounded | [xs:string](#38) |  |
| **Data Standard Version (53794)** | The data standard version that the finding and coding corresponds to i.e. 21.0.0 RF2 | 0..unbounded | [xs:string](#38) |  |
| **Qualifier Type ID (55340)** | Identifier for the qualifier type | 1..1 | [xs:string](#38) |  |
| **Qualifier Type (55338)** | Name of the qualifier | 1..1 | [xs:string](#38) |  |
| **Qualifier (55339)** | Qualifier | 1..1 | [xs:string](#38) |  |

## Death

This section is used to submit details of the date and cause of death. Participants remain in the programme after their death but this information is crucial both for project implementation (to ensure appropriate future contact with the family) and for research. All fetal participants should also have a death details form completed at the time of recruitment; this is because samples from ongoing pregnancies are not eligible for the programme, as genome sequencing is not yet fast enough to be used in the context of ongoing pregnancy. The date of death should refer to the date the intra-uterine death was discovered, or feticide was carried out. 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** | **Related To** |
| **Death Location (12777)** | Location of death | 0..1 | [deathLocation](#54202) **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 |  |
| **Significant (12781)** | Significant condition not 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..\* | [deathCauseCode](#54201) |  |
| **Immediate Cause (12778)** | 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. | 1..\* | [deathCauseCode](#54201) |  |
| **Condition (12780)** | 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..\* | [deathCauseCode](#54201) |  |
| **Underlying Cause (12779)** | 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..\* | [deathCauseCode](#54201) |  |

### Participant Identifiers [1..1]

*One report containing Participant Identifiers must be submitted together with each Death report.*

The following information is used to identify the participant and must be included with all data submissions.  
For non-NHSE participants, including overseas participants, who do not have an NHS number, the dummy NHS number 2222222222 should be used.  
For fetal participants: the expected due date should be entered in place of the date of birth. Forename should be entered as ‘Fetus of mother’s forename’, for example ‘Fetus of Anna’. The mother’s surname should be used for the surname. The dummy NHS number 333333333 should be used. A death form should be recorded at the time of registration for deceased fetuses.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Participant ID (12502)** | Participant Identifier (supplied by Genomics England) | 1..1 | [participantId](#54269) |  |
| **Date of Birth (12505)** | The date on which a PERSON was born or is officially deemed to have been born. | 1..1 | [xs:date](#58) | PERSON BIRTH DATE (NHS Data Dictionary GEL Subset) |
| **Surname (12507)** | The participant's surname | 1..1 | [personFamilyName](#54276) | PERSON FAMILY NAME (CR0050 from Cancer Outcomes and Services Dataset) |
| **Forenames (12508)** | The participant's forenames | 1..1 | [personGivenName](#54278) | PERSON GIVEN NAME (CR0060 from Cancer Outcomes and Services Dataset) |

#### Person Identifier [1..1]

*One report containing Person Identifier must be submitted together with each Participant Identifiers report.*  
  
Choice of one of either NHS Number (Wales & England) OR CHI Number (Scotland) OR Health and Care Number (Northern Ireland). Patients without an NHS number (or equivalent) should be allocated a temporary NHS number by the treating hospital.

*One of the following must be submitted together with each Person Identifier report.*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **NHS Number (12506@1.0.1)** | Validated NHS number for participant | 1..1 | [nhsNumber](#42014) |  |
| **Or in the case of families recruited to the Scottish Genomes Project,** | | | | |
| **CHI Number (14821@1.0.1)** | The COMMUNITY HEALTH INDEX NUMBER (CHI NUMBER) uniquely identifies a PATIENT on the Community Health Index (Scotland) within the NHS in Scotland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [chiNumber](#42011) |  |
| **Or in the case of families recruited by the Northern Ireland Genomic Medicine Centre,** | | | | |
| **Health and Care Number (42126@1.0.1)** | Validated HEALTH AND CARE NUMBER (H&C NUMBER). Uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales. | 1..1 | [healthAndCareNumber](#42127) |  |

### Event Details [1..1]

*One report containing Event Details must be submitted together with each Death report.*

The local ‘Event Reference’ combined with the Event Date, is used to identify the actual participant event involved. GMCs must ensure that local event references are not re-used to refer to different events, therefore two different events on the same date can be easily identified. GMCs must also ensure no two participants within their GMC are given the same event reference.  
  
If a second submission is received with the same local event reference, then Genomics England will assume that this is an update pertaining to the same participant event, and will be used in place of the original submission; any data values previously supplied that are still applicable must therefore be included.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Name** | **Description** | **Multiplicity** | **Data Type** | **Related To** |
| **Event Date (12727)** | Date of the clinical event or observation being reported e.g. date biopsy was taken | 1..1 | [xs:dateTime](#59) |  |
| **Event Reference (14858)** | Unique identifier for local record of clinical event or observation | 1..1 | [xs:string](#38) |  |

# Additional Data [0..\*]

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.

# **Data Types**

**ICD10Code**

**(Rare Diseases)**

Character strings in XML.

|  |  |
| --- | --- |
| Rule based on  icdCode (Genomics England Shared 1.2.0) | maxLength(6) |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Medical Condition](#39092)

**MultipleMonogenicCauses**

**(Rare Diseases)**

Multiple Monogenic Causes:   
yes- segregating  
yes: segregating separately  
No

|  |  |
| --- | --- |
| **Code** | **Description** |
| Segregating | Segregaing |
| Segregating\_separately | Segregating Separately |
| No | No |

**Usages**

[Proband](#34016)

**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

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

**Usages**

[Participant Contact Details](#54263)

**Present**

**(Rare Diseases)**

Presence of the phenotypic abnormality.

|  |  |
| --- | --- |
| Based On | Phenotype\_Present (Rare Diseases 2.0.0) |
| **Code** | **Description** |
| unknown | Unknown |
| yes | Yes |
| no | No |

**Usages**

[Phenotype Statement](#33959)

**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

|  |  |
| --- | --- |
| Regular Expression | .{2,175} |

**Usages**

[Participant Contact Details](#54263)

**adoptedStatus**

**(Rare Diseases)**

|  |  |
| --- | --- |
| **Code** | **Description** |
| not\_adopted | not adopted |
| adopted\_in | adopted into the family |
| adopted\_out | child belonged to the family and was adopted out |

**Usages**

[Pedigree Member](#33940)

**affectedDiseaseStatus**

**(Rare Diseases)**

|  |  |
| --- | --- |
| **Code** | **Description** |
| affected | Affected |
| unaffected | Unaffected |
| uncertain | Uncertain |

**Usages**

[Pedigree Member](#33940)

**affectedStatus**

**(Rare Diseases)**

Yes / no / unknown

|  |  |
| --- | --- |
| Based On | yesNoUnk (Genomics England Shared 0.0.9) |
| Based On | yesNoUnk (Genomics England Shared 1.2.0) |
| **Code** | **Description** |
| yes | Yes |
| no | No |
| unknown | Unknown |

**Usages**

[Proband](#34016)

**ageInYearsFractions**

**(Rare Diseases)**

Age in years as a fraction

|  |  |
| --- | --- |
| Unit of Measure | Year (A year is the orbital period of the Earth moving in its orbit around the Sun |
| Rule based on  xs:double (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  parseDouble(string(x)) in Double |

**Usages**

[Disease Information GEL](#33370)

[Disease Information Other](#33371)

[Pedigree Member](#33940)

**biologicalFamilyRelationship**

**(Rare Diseases)**

Biological Family Relationship

|  |  |
| --- | --- |
| **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 |
| DoubleFirstCousin | Double First Cousin |
| Son | Son |
| Daughter | Daughter |
| Other | Other (Please specify) |
| MaternalGrandparent | Mother’s parent |
| PaternalGrandparent | Father’s parent |
| MaternalGrandfatherParent | Maternal great-grandparent through grandfather - parent of maternal grandfather |
| MaternalGrandmotherParent | Maternal great-grandparent through grandmother - parent of maternal grandmother |
| PaternalGrandfatherParent | Paternal great-grandparent through grandfather - parent of paternal grandfather |
| PaternalGrandmotherParent | Paternal great-grandparent through grandmother - parent of paternal grandmother |
| MaternalFirstCousinOnceRemoved | Child of proband's maternal first cousin or first cousin of proband's mother |
| PaternalFirstCousinOnceRemoved | Child of proband's paternal first cousin or first cousin of proband's father |
| MaternalSecondCousin | Maternal second cousin (shared set of great-grandparents to which proband is related through mother) |
| PaternalSecondCousin | Paternal second cousin (shared set of great-grandparents to which proband is related through father) |
| MaternalSecondCousinOnceRemoved | Child of proband's maternal second cousin or second cousin of proband's mother |
| PaternalSecondCousinOnceRemoved | Child of proband's paternal second cousin or second cousin of proband's father |
| MaternalThirdCousin | Maternal third cousin (shared set of great-great-grandparents to which proband is related through mother) |
| PaternalThirdCousin | Paternal third cousin (shared set of great-great-grandparents to which proband is related through father) |

**Usages**

[Relative](#34073)

**chiNumber**

**(NHS Data Dictionary GEL Subset)**

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.

|  |  |
| --- | --- |
| Regular Expression | [a-zA-Z0-9]{10} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Person Identifier](#54271)

**clinicalTestAbnormality**

**(Rare Diseases)**

Clinical Test Abnormality

|  |  |
| --- | --- |
| **Code** | **Description** |
| 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 |

**Usages**

[Finding](#53804)

[Genetic Results](#33494)

[Laboratory Test](#53802)

**consanguinity**

**(Rare Diseases)**

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

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | YesNoUnkPos (Rare Diseases 2.0.0) | | |
| **Code** | | **Description** |
| U | | Unknown |
| P | | Possible |
| N | | No |
| Y | | Yes |

**Usages**

[Pedigree Member](#33940)

[Pedigree Member Relationship](#55355)

[Proband](#34016)

**consentWithdrawalOptions**

**(Genomics England Shared)**

Genomics England Consent Withdrawal Options

|  |  |
| --- | --- |
| **Code** | **Description** |
| full\_withdrawal | OPTION 2: FULL WITHDRAWAL: No further use |
| partial\_withdrawal | OPTION 1: PARTIAL WITHDRAWAL: No further contact |

**Usages**

[Withdrawal](#54314)

**consultantCode**

**(Cancer Outcomes and Services Dataset)**

The GMC code of the consultant

|  |  |
| --- | --- |
| Rule | x==~/[a-zA-Z0-9]{1,8}/ |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Consultant Details](#54193)

**deathCauseCode**

**(Genomics England Shared)**

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

|  |  |
| --- | --- |
| Regular Expression based on  deathCauseIcdCode (Cancer Outcomes and Services Dataset 6.0.0) | [a-zA-Z0-9.]{3,6} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Death](#54197)

**deathLocation**

**(Genomics England Shared)**

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.

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | deathLocationType (NHS Data Dictionary GEL Subset 1.0.0) | | |
| **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 |

**Usages**

[Death](#54197)

**diagnosticTermsSnomedCT**

**(Rare Diseases)**

SNOMED CT CODE

|  |  |
| --- | --- |
| Regular Expression based on  snomedCt (Genomics England Shared 1.2.0) | \d{6,18} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Medical Condition](#39092)

**emailAddress**

**(Genomics England Shared)**

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

|  |  |
| --- | --- |
| Rule | x ==~/(?:[a-z0-9!#$%&'\*+\/=?^\_`{|}~-]+(?:\.[a-z0-9!#$%&'\*+\/=?^\_`{|}~-]+)\*|"(?:[\x01-\x08\x0b\x0c\x0e-\x1f\x21\x23-\x5b\x5d-\x7f]|\\[\x01-\x09\x0b\x0c\x0e-\x7f])\*")@(?:(?:[a-z0-9](?:[a-z0-9-]\*[a-z0-9])?\.)+[a-z0-9](?:[a-z0-9-]\*[a-z0-9])?|\[(?:(?:25[0-5]|2[0-4][0-9]|[01]?[0-9][0-9]?)\.){3}(?:25[0-5]|2[0-4][0-9]|[01]?[0-9][0-9]?|[a-z0-9-]\*[a-z0-9]:(?:[\x01-\x08\x0b\x0c\x0e-\x1f\x21-\x5a\x53-\x7f]|\\[\x01-\x09\x0b\x0c\x0e-\x7f])+)\])/ |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Participant Contact Details](#54263)

**ethnicCategory**

**(Genomics England Shared)**

|  |  |
| --- | --- |
| **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 |
| 99 | Not known |

**Usages**

[Family History](#39087)

[Pedigree Member](#33940)

[Registration](#33925)

**geneScope**

**(Genomics England Shared)**

The gene or genes considered

|  |  |
| --- | --- |
| Based On | hgncSymbol (Genomics England Shared 1.2.0) |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Genetic Result](#54217)

**geneticTestLaboratory**

**(Genomics England Shared)**

Was genetic testing performed in a diagnostic or research laboratory?

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | genetic\_test\_laboratory (Genomics England Shared 1.2.0) | | |
| **Code** | | **Description** |
| research\_laboratory | | Research laboratory |
| diagnostic\_laboratory | | Diagnostic laboratory |

**Usages**

[Genetic Result](#54217)

**geneticTestMethod**

**(Genomics England Shared)**

Genetic Test Method

|  |  |
| --- | --- |
| **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 | Other SNV analysis e.g. OLA |
| targeted\_copy\_number\_analysis | 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\_breakage\_testing | Fanconi (MMC/DEB) breakage testing |
| radiation\_hypersensitivity | 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 | Translocation eg qPCR/sequencing/FISH/IHC |
| other | Other |

**Usages**

[Genetic Result](#54217)

**genomicsEnglandConsentWithdrawalForms**

**(Genomics England Shared)**

List of consent withdrawal forms used by Genomics England

|  |  |
| --- | --- |
| **Code** | **Description** |
| 6a | Withdrawal information and form – for adult or child participants (6a) |
| 6b | Consultee declaration of advice regarding adult participant withdrawal information – for consultees (withdrawal) (6b) |

**Usages**

[Withdrawal](#54314)

**gmcFamilyId**

**(Rare Diseases)**

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

|  |  |
| --- | --- |
| Regular Expression | [0-9]{9} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Pedigree Data](#33938)

[Registration](#33925)

**groupType**

**(Rare Diseases)**

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.

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | TrioDuoSingletonTypes (Rare Diseases 2.0.0) | | |
| **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 |

**Usages**

[Proband](#34016)

**healthAndCareNumber**

**(Genomics England Shared)**

The HEALTH AND CARE NUMBER (H&C NUMBER) uniquely identifies a PATIENT within the NHS in Northern Ireland. It is the equivalent of the NHS NUMBER in England and Wales.  
  
The HEALTH AND CARE NUMBER is ten numeric digits in length, and is in the same format as the NHS NUMBER in England (3 3 4 format with the tenth digit being a modulus 11 check digit). HEALTH AND CARE NUMBERS are however unique from NHS NUMBERS allocated in England as they are taken from a range of numbers reserved for Northern Ireland (320 000 001 to 399 999 999 plus check digit).

|  |  |
| --- | --- |
| Regular Expression | [a-zA-Z0-9]{10} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Person Identifier](#54271)

**heredityStatus**

**(Rare Diseases)**

|  |  |
| --- | --- |
| **Code** | **Description** |
| none | None |
| infertile | Infertile |
| childless | Childless |

**Usages**

[Pedigree Member](#33940)

**hpoID**

**(Rare Diseases)**

The identity of the statement within the Human Phenotype Ontology

|  |  |
| --- | --- |
| Regular Expression | HP:[0-9]{7} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Modifier](#55367)

[Phenotype Statement](#33959)

**lifeStatus**

**(Rare Diseases)**

|  |  |
| --- | --- |
| **Code** | **Description** |
| alive | alive |
| aborted | aborted |
| deceased | deceased |
| unborn | unborn |
| stillborn | stillborn |
| miscarriage | miscarriage |

**Usages**

[Pedigree Member](#33940)

**molecularTestResult**

**(Genomics England Shared)**

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'.

|  |  |
| --- | --- |
| **Code** | **Description** |
| normal | Normal (negative) |
| fail | Fail |
| abnormalitydetected | Pathogenic abnormality detected |
| vus | Variant of unknown significance detected |

**Usages**

[Genetic Result](#54217)

**nhsNumber**

**(NHS Data Dictionary GEL Subset)**

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.

|  |  |
| --- | --- |
| 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 |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Couple Report Details](#39079)

[Person Identifier](#54271)

**organisationSiteCode**

**(NHS Data Dictionary GEL Subset)**

An Organisation site code or ODS code identifies an NHS Organisation uniquely

|  |  |
| --- | --- |
| Regular Expression | [a-zA-Z0-9]{3,9} |
| Rule based on  organisationSiteCode (Cancer Outcomes and Services Dataset 6.0.0) | x ==~/[a-zA-Z0-9]{3,9}/ |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Consultant Details](#54193)

[Registration](#33925)

**participantId**

**(Genomics England Shared)**

Genomics England participant identifier (supplied by Genomics England)

|  |  |
| --- | --- |
| Regular Expression | \d{9} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Couple Report Details](#39079)

[Participant Identifiers](#54267)

[Pedigree Member](#33940)

**participantType**

**(Rare Diseases)**

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

|  |  |
| --- | --- |
| **Code** | **Description** |
| Proband | Proband |
| Relative | Relative |

**Usages**

[Clinical Information](#33276)

[Pedigree Member](#33940)

**patientStatus**

**(Rare Diseases)**

Represents the patient's status when test was performed

|  |  |
| --- | --- |
| **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 |

**Usages**

[Finding](#53804)

[Laboratory Test](#53802)

**penetrance**

**(Rare Diseases)**

Yes / no / unknown

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | yesNoUnk (Genomics England Shared 0.0.9) | | |
| Based On | yesNoUnk (Genomics England Shared 1.2.0) | | |
| **Code** | | **Description** |
| yes | | Yes |
| no | | No |
| unknown | | Unknown |

**Usages**

[Proband](#34016)

**personFamilyName**

**(Genomics England Shared)**

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

|  |  |
| --- | --- |
| Regular Expression based on  personName (Genomics England Shared 1.2.0) | [a-zA-Z0-9\s-']{1,50} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Participant Identifiers](#54267)

**personFamilyNameAtBirth**

**(Genomics England Shared)**

The PATIENT's surname at birth.

|  |  |
| --- | --- |
| Regular Expression based on  personName (Genomics England Shared 1.2.0) | [a-zA-Z0-9\s-']{1,50} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Registration](#33925)

**personGivenName**

**(Genomics England Shared)**

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

|  |  |
| --- | --- |
| Regular Expression based on  personName (Genomics England Shared 1.2.0) | [a-zA-Z0-9\s-']{1,50} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Participant Identifiers](#54267)

**personKaryotypicSexClassification**

**(Rare Diseases)**

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | personKaryotypicSex (Rare Diseases 2.0.0) | | |
| **Code** | | **Description** |
| XY | | XY |
| XX | | XX |
| XO | | XO |
| XXY | | XXY |
| XYY | | XYY |
| XXX | | XXX |
| XXYY | | XXYY |
| XXXY | | XXXY |
| XXXX | | XXXX |
| other | | other |
| unknown | | unknown |

**Usages**

[Pedigree Member](#33940)

[Registration](#33925)

**personPhenotypicSexClassification**

**(NHS Data Dictionary GEL Subset)**

A classification of PERSON PHENOTYPIC SEX  
http://www.datadictionary.nhs.uk/data\_dictionary/attributes/p/person/person\_phenotypic\_sex\_classification\_de.asp?shownav=1

|  |  |  |  |
| --- | --- | --- | --- |
| Based On | personPhenotypicSex (Genomics England Shared 1.0.0) | | |
| Based On | personPhenotypicSex (Genomics England Shared 1.0.1) | | |
| **Code** | | **Description** |
| 2 | | Female |
| 1 | | Male |
| 9 | | Indeterminate |

**Usages**

[Pedigree Member](#33940)

[Registration](#33925)

**personStatedGenderCode**

**(Genomics England Shared)**

The participant's current gender. COSD v7 update

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

**Usages**

[Registration](#33925)

**positiveInteger**

**(Rare Diseases)**

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

|  |  |
| --- | --- |
| Rule | minInclusive(0) |
| Rule based on  xs:integer (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  parseInteger(string(x)) in BigInteger |

**Usages**

[Proband](#34016)

**samplingPreconditions**

**(Rare Diseases)**

|  |  |
| --- | --- |
| **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 |

**Usages**

[Laboratory Test](#53802)

**specialty**

**(Cancer Model)**

The specialty code of the person performing the event

|  |  |
| --- | --- |
| Regular Expression | [a-zA-Z0-9]{3} |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Procedure](#55341)

**testStatus**

**(Rare Diseases)**

event status

|  |  |
| --- | --- |
| **Code** | **Description** |
| not\_done | Not done |
| failed | Failed |
| complete | Complete |
| pending | Pending |

**Usages**

[Laboratory Test](#53802)

**timeAspect**

**(Rare Diseases)**

|  |  |
| --- | --- |
| **Code** | **Description** |
| Spot | Spot |
| Unspecified | Unspecified |
| 24hr Collection | 24hr Collection |
| Basal | Basal - Time Course |
| Stimulated Peak | Stimulated Peak - Time Course |

**Usages**

[Laboratory Test](#53802)

**tissueOrigin**

**(Rare Diseases)**

Origin of the tissue

|  |  |
| --- | --- |
| **Code** | **Description** |
| Blood | Blood |
| Skin | Skin |
| Saliva | Saliva |
| Muscle | Muscle |
| CVS | CVS |
| Amniocentesis | Amniocentesis |
| Tumour | Tumour |
| Other | Other |

**Usages**

[Genetic Results](#33494)

**treatmentIntent**

**(Genomics England Shared)**

Intent of the proposed treatment

|  |  |
| --- | --- |
| **Code** | **Description** |
| C | Curative |
| Z | Non Curative |
| X | No active treatment |
| 9 | Not Known |

**Usages**

[Therapy Regime](#33800)

**ukTelephoneNumber**

**(Genomics England Shared)**

uk phone number

|  |  |
| --- | --- |
| Regular Expression | ^(((\+44\s?\d{4}|\(?0\d{4}\)?)\s?\d{3}\s?\d{3})|((\+44\s?\d{3}|\(?0\d{3}\)?)\s?\d{3}\s?\d{4})|((\+44\s?\d{2}|\(?0\d{2}\)?)\s?\d{4}\s?\d{4}))(\s?\#(\d{3,5}))?$ |
| Rule based on  xs:string (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Consultant Details](#54193)

[Participant Contact Details](#54263)

**xs:boolean**

**(XMLSchema)**

Binary-valued logic legal literals

|  |  |
| --- | --- |
| **Code** | **Description** |
| 0 | False |
| 1 | True |
| true | True |
| false | False |

**Usages**

[Pedigree Member](#33940)

[Pedigree Member Relationship](#55355)

[Procedure](#55341)

**xs:date**

**(XMLSchema)**

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 |

**Usages**

[Couple Report Details](#39079)

[Participant Identifiers](#54267)

[Pedigree Member](#33940)

[Relative](#34073)

[Sample Details](#51969)

**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.

|  |  |
| --- | --- |
| Rule | import static javax.xml.bind.DatatypeConverter.\*  parseDateTime(string(x)) in Calendar |

**Usages**

[Event Details](#54208)

[Finding](#53804)

[Laboratory Test](#53802)

[Procedure](#55341)

**xs:duration**

**(XMLSchema)**

A duration of time. ISO 8601 extended format PnYnMnDTnHnMnS. Example, to indicate duration of 1 year, 2 months, 3 days, 10 hours, and 30 minutes: P1Y2M3DT10H30M. One could also indicate a duration of minus 120 days as: -P120D.

|  |  |
| --- | --- |
| Regular Expression | -?P\d+Y(\d+M(\d+D(T\d+H(\d+M(\d+S)?)?)?)?)? |

**Usages**

[Finding](#53804)

[Laboratory Test](#53802)

[Procedure](#55341)

**xs:integer**

**(XMLSchema)**

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

|  |  |
| --- | --- |
| Rule | import static javax.xml.bind.DatatypeConverter.\*  parseInteger(string(x)) in BigInteger |

**Usages**

[Pedigree Data](#33938)

[Pedigree Member](#33940)

[Pedigree Member Relationship](#55355)

**xs:positiveInteger**

**(XMLSchema)**

Infinite set {1, 2,...}. Optional “+” sign,. Example: 1, 12678967543233, +100000.

|  |  |
| --- | --- |
| Rule | minExclusive(0) |
| Rule based on  xs:nonNegativeInteger (XMLSchema 0.0.1) | minInclusive(0) |
| Rule based on  xs:integer (XMLSchema 0.0.1) | import static javax.xml.bind.DatatypeConverter.\*  parseInteger(string(x)) in BigInteger |

**Usages**

[Proband](#34016)

**xs:string**

**(XMLSchema)**

Character strings in XML.

|  |  |
| --- | --- |
| Rule | import static javax.xml.bind.DatatypeConverter.\*  true && (x = parseString(string(x))) |

**Usages**

[Consent](#39093)

[Consent Update](#39100)

[Consultant Details](#54193)

[Couple Report Details](#39079)

[Disease Information GEL](#33370)

[Eligibility](#33403)

[Event Details](#54208)

[Family History](#39087)

[Finding](#53804)

[Genetic Reports](#33496)

[Genetic Result](#54217)

[Genetic Results](#33494)

[Laboratory Test](#53802)

[Medical Condition](#39092)

[Modifier](#55367)

[Pedigree Data](#33938)

[Pedigree Member](#33940)

[Phenotype Report](#33957)

[Phenotype Statement](#33959)

[Procedure](#55341)

[Qualifier](#55336)

[Registration](#33925)

[Relative](#34073)

[Sample Details](#51969)

[Therapy Regime](#33800)

[Withdrawal](#54314)

**yesNo**

**(Genomics England Shared)**

Boolean, yes no response

|  |  |
| --- | --- |
| **Code** | **Description** |
| yes | Yes |
| no | No |

**Usages**

[Consent](#39093)

[Consent Details](#55330)

[Consent Update](#39100)

[Eligibility](#33403)

[Pedigree Member](#33940)

[Proband](#34016)

[Registration](#33925)

**yesNoNotRelevant**

**(Genomics England Shared)**

yes, no, not relevant

|  |  |
| --- | --- |
| **Code** | **Description** |
| yes | yes |
| no | no |
| not\_relevant | not relevant |

**Usages**

[Consent Details](#55330)

**yesNoUnk**

**(Genomics England Shared)**

Yes / no / unknown

|  |  |
| --- | --- |
| **Code** | **Description** |
| yes | Yes |
| no | No |
| unknown | Unknown |

**Usages**

[Pedigree Member](#33940)

# Validation Rules

Disease group, subgroup Disease Group, Subgroup and Specific disease consistency.

|  |  |
| --- | --- |
| Rule | **Disease group, subgroup Disease Group, Subgroup and Specific disease must be consistent with level 2, level 3 and level 4 disorders in Appendix A Genomics England Rare Disease List.** |
| Last Updated | **2017-02-27** |
| Version Created | **2017-02-27** |
| Status | **DRAFT** |

Participants in family and group type should be consistent

|  |  |
| --- | --- |
| Rule | **Participants in family and group type should be consistent i.e. if group is a trio then family size must be 3** |
| Last Updated | **2017-02-24** |
| Version Created | **2017-02-24** |
| Status | **DRAFT** |