

# FAIR GENOMES: Join us in standardizing meta-data for personal genome workflows all over the Netherlands

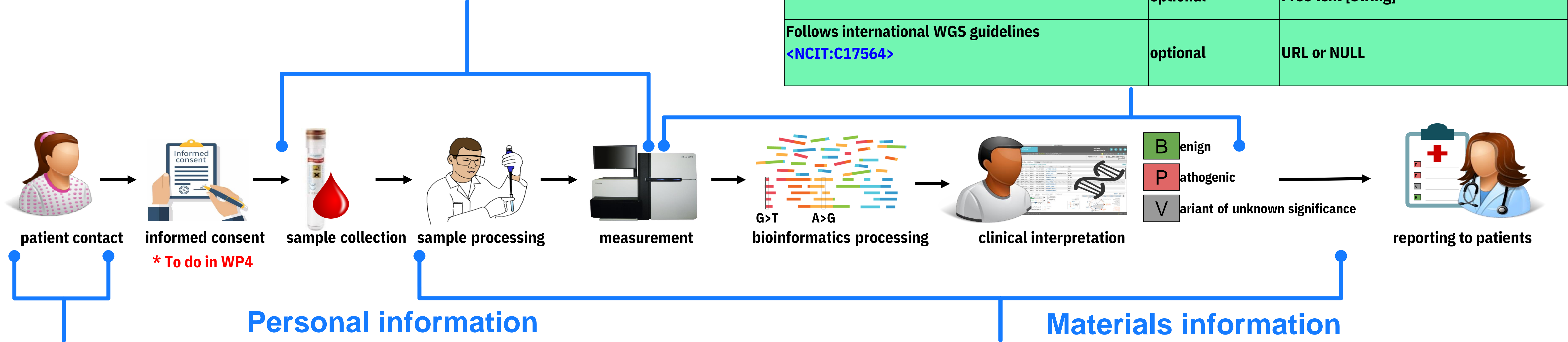


## Clinical information

Meta-data element	Compulsory / optional	Value types
Phenotypic terms <MESH:D010641>	compulsory	Ontological terms based on <b>HPO / ORDO</b>
Unobserved phenotypes <HL7:C0442737>	optional	Ontological terms based on <b>HPO / ORDO</b>
Type of phenotypic data <DC:DCMIType>	compulsory	Ontological terms subclass of <DC:DCMIType>
Clinical diagnosis <SNOMEDCT:39154008>	optional	Ontological terms based on <b>ORDO / ICD10 / SNOMED</b>
Genetic diagnosis <CRISP:1254-8491>	optional	Identifiers based on <b>OMIM gene, HGNC gene, variant in HGVS</b>
Age at diagnosis <SNOMEDCT:423493009>	optional	YYYY
Age at last screening <NCIT:C81258>	optional	YYYY
Medication information <EDAM:data_3103>	optional	Ontological terms based of <b>ATC codes</b>
Dosage	optional	Free text [String]
Family members affected	optional	Phenopakket buildingblock
Family members sequenced <UMLS:C0086282>	optional	Phenopackets buildingblock
Procedural history <UMLS:C1138431 >	optional	Ontological terms based on <b>CPT ontology</b>
Age of onset <HP:0003674>	optional	[Positive Integer]

## Technical information

Meta-data element	Compulsory / optional	Value types
Sequencing date <GENEPIO:0000069>	Compulsory	YYYY-MM-DD HH-MM-SS
Sample prep kit <GENEPIO:0000081>	Optional	Free text [String]   Ontological terms subclass of <BAO:0002091>
Sequencing platform <GENEPIO:0000071>	compulsory	Free text [String]   Ontological terms subclass of <GENEPIO:0000071>
Sequencing data type <NCIT:C18881>	compulsory	Ontological terms subclass of <NCIT:C18881>
PCR-free <NCIT:C17003>	optional	[Boolean]
Sequencing average read depth <NCIT:C155320>	compulsory	[Positive Integer]
Enrichment panel used <NCIT:C154307>	compulsory	Free text [String]   Ontological terms subclass of <BAO:0002091>
UMIs present <EFO:0010199>	optional	[Boolean]
Read length <NCIT:C153362>	compulsory	[Positive Integer]
insert size	optional	[Positive Integer]
Location of data <DUO:GAZ_00000448>	compulsory	Free text [String]
Type of data stored <EDAM:FORMAT_2350>	compulsory	Ontological terms subclass of <EDAM:FORMAT_2350>
Used algorithms <NCIT:C16275>	optional	Free text [String]
Bioinformatics analysis protocol<EDAM:DATA_2531>	optional	URL
Special algorithms parameters used <NCIT:C44175>	optional	Free text [String]
Follows international WGS guidelines <NCIT:C17564>	optional	URL or NULL



## Personal information

Meta-data element	Compulsory / optional	Value types
Patient ID <SDO:PatientID>	compulsory	ID [String]
Biological sex <SIO:010029>	compulsory	1. Male <SIO:010048> 2. Female <SIO:010052> 3. NULL <hl7:v3-NullFlavor>
Country of residence <SNOMEDCT:276205001>	optional	Ontological term subclass of <SNOMEDCT:370159000>
Ethnicity <LOINC:MTHU010275>	optional	Ontological term subclass of <SNOMEDCT:370159000>
Country of birth <SNOMEDCT:370159000>	optional	Ontological term subclass of <SNOMEDCT:370159000>
Year of birth <NCIT:C83164>	optional	YYYY (4 digits, Gregorian)
Patient status <NCIT:C166244>	optional	1. Alive <NCIT:C37987> 2. Dead <NCIT:C28554> 3. Lost in follow-up <NCIT:C48227> 4. Opted-out <HL7:C3858493>
Age at death <GSSO:C135383>	optional	YYYY (4 digits, Gregorian)
Inclusion criterion <OBIB:0500027>	optional	Free text [STRING]

## Materials information

Meta-data element	Compulsory / optional	Value types
Sampling time stamp <UMLS:C0870078>	compulsory	YYYY-MM-DD HH-MM-SS
Registration time stamp <CTO:Registration>	compulsory	YYYY-MM-DD HH-MM-SS
Sampling collection protocol <EFO:0005518>	optional	Free text [String]
Deviations from sample protocol <NCIT:C50996>	optional	Free text [String]
Reasons for protocol deviation	optional	Free Text [String]
Material type <UMLS:C2986062>	compulsory	Ontological terms based on <b>NCIT / PT / Material Identifier Type code</b>
Anatomical source <UBERON:0001062>	optional	Ontological terms based on <b>UBERON</b> code
Storage conditions <UMLS:C3272596>	optional	Ontological terms based on <b>OBIB</b> ontology
Expiration date <LOINC:C3669020>	optional	YYYY-MM-DD
Estimated percentage of tumor cells <UMLS:C4288090>	optional	0-100 [Integer]

Contribute now or contribute later! Scan the QR code to retrieve the latest schema



“FAIR genomes: a national guideline to promote optimal (re)use of NGS data in research and healthcare” is a ZonMw project in the Personalised Medicine programme registered under project number 846003201.

