Clinical data dictionary

175 fields in 17 sections, with types, allowed values and missing-data rules.

All fields of release 1.0. “Repeatable” sections hold a list of entries. Empty value = not recorded. Dates are ISO 8601 (YYYY-MM-DD) in exports. Numbers keep the unit recorded with them.

Status value	Meaning (missing-data rules)
Present	Finding was assessed and is present.
Absent	Finding was explicitly assessed and is not present. Exported as an excluded phenotype.
Suspected	Finding is possible but not confirmed. Kept apart from confirmed findings; left out of ranking unless switched on.
Unknown	Asked or looked for, result not known.
Not assessed	Not examined or asked.
Not applicable	Cannot apply to this patient (for example by age or sex).
(no record)	Nothing was recorded. Never treated as absent.

Registration and demographics

Field	Label	Type	Allowed values
reg.hospitalId	Hospital / institution ID	text	
reg.studyId	Study / cohort ID	text	
reg.first	First name	text	
reg.last	Last name	text	
reg.dob	Date of birth	date	
reg.sex	Biological sex	choice	Female | Male | Intersex | Unknown
reg.gender	Gender identity (optional)	text	
reg.ancestry	Ancestry (optional)	text	
reg.country	Country	text	
reg.location	City / region	text	
reg.phone	Phone	text	
reg.email	Email	email	
reg.refPhys	Referring physician	text	

reg.geneticist	Clinical geneticist	text	
reg.clinician	Primary clinician	text	
reg.regDate	Registration date	date	
reg.caseType	Case type	choice	Adult | Paediatric | Neonatal | Prenatal | Deceased
reg.familyId	Family ID (family-based study)	text	
reg.dataStatus	Data collection status	choice	Not started | In progress | Complete | On hold
reg.dxStatus	Diagnostic status	choice	Undiagnosed | Clinical diagnosis | Molecular diagnosis | Partially explained
reg.indication	Referral indication	long text	

Presenting complaint (repeatable)

Field	Label	Type	Allowed values
complaints.kind	Type	choice	Primary | Secondary
complaints.desc	Clinical description	long text	
complaints.onset	Age / date of onset	text	
complaints.duration	Duration	text	
complaints.progression	Progression	choice	Stable | Progressive | Improving | Episodic | Resolved
complaints.severity	Severity	choice	Borderline | Mild | Moderate | Severe | Profound
complaints.frequency	Frequency	choice	Constant | Frequent | Occasional | Rare | Single episode
complaints.associated	Associated manifestations	text	
complaints.triggers	Triggering / alleviating factors	text	
complaints.prevDx	Previous diagnoses	text	
complaints.source	Source of information	choice	Clinician-observed | Patient-reported | Caregiver-reported | Documented in a medical record | Derived from a test

Medical history

Field	Label	Type	Allowed values
med.diseases	Previous diseases	long text	
med.chronic	Chronic conditions	long text	
med.hospital	Hospitalisations	long text	

med.surgery	Surgeries	long text	
med.prevDx	Previous diagnoses	long text	
med.prevGenetic	Previous genetic testing	long text	
med.allergies	Allergies	long text	
med.adr	Adverse drug reactions	long text	

Medication (repeatable)

Field	Label	Type	Allowed values
meds.name	Medication	text	
meds.dose	Dose and schedule	text	
meds.start	Start	date	
meds.stop	Stop	date	
meds.response	Treatment response	choice	Good | Partial | None | Worsened | Unknown
meds.adr	Adverse reaction	text	

Birth and developmental history

Field	Label	Type	Allowed values
birth.pregnancy	Pregnancy history	long text	
birth.maternal	Maternal complications	long text	
birth.gestWeeks	Gestational age (weeks)	number	
birth.delivery	Delivery type	choice	Vaginal | Assisted vaginal | Elective caesarean | Emergency caesarean | Unknown
birth.bw	Birth weight (g)	number	
birth.bl	Birth length (cm)	number	
birth.ofc	Head circumference at birth (cm)	number	
birth.apgar1	APGAR 1 min	number	
birth.apgar5	APGAR 5 min	number	
birth.nicu	NICU admission	choice	Yes | No | Unknown
birth.neonatal	Neonatal complications	long text	
birth.congenital	Congenital abnormalities	long text	
birth.sat	Sat unsupported (months)	number	
birth.walked	Walked (months)	number	

birth.words	First words (months)	number	
birth.regression	Developmental regression	choice	Yes | No | Unknown
birth.intellectual	Intellectual development	long text	
birth.speech	Speech / language development	long text	
birth.motor	Motor development	long text	
birth.behaviour	Behavioural development	long text	

Family history (family level)

Field	Label	Type	Allowed values
famx.consang	Consanguinity	choice	Yes | No | Unknown
famx.consangDetail	Consanguinity detail	text	
famx.miscarriages	Miscarriages	text	
famx.stillbirths	Stillbirths	text	
famx.infantDeaths	Infant deaths	text	
famx.knownDisorders	Known genetic disorders in the family	long text	
famx.parentalAges	Parental ages at birth	text	
famx.ancestryNote	Relevant ancestry	text	

Relative (repeatable)

Field	Label	Type	Allowed values
member.rel	Relationship	choice	Mother | Father | Brother | Sister | Sibling (sex unknown) | Maternal grandmother | Maternal grandfather | Paternal grandmother | Paternal grandfather | Maternal aunt | Maternal uncle | Paternal aunt | Paternal uncle | Son | Daughter | Partner | Other / uncertain
member.label	Name or label	text	
member.sex	Sex	choice	Female | Male | Unknown
member.vital	Vital status	choice	Alive | Deceased | Unknown
member.age	Age / age at death	text	
member.affected	Affected	choice	Yes | No | Unknown
member.phenotype	Phenotype	text	
member.onset	Age of onset	text	

member.tested	Testing status	choice	Not tested | Tested – positive | Tested – negative | Pending | Unknown
member.notes	Notes	text	

Phenotype observation (repeatable)

Field	Label	Type	Allowed values
obs.status	Status	choice	Present | Absent | Suspected | Unknown | Not assessed | Not applicable
obs.source	Evidence source	choice	Clinician-observed | Patient-reported | Caregiver-reported | Documented in a medical record | Derived from a test
obs.onset	Age / date of onset	text	
obs.resolved	Resolution (age / date)	text	
obs.severity	Severity	choice	Borderline | Mild | Moderate | Severe | Profound
obs.progression	Progression	choice	Stable | Progressive | Improving | Episodic | Resolved
obs.frequency	Frequency	choice	Constant | Frequent | Occasional | Rare | Single episode
obs.laterality	Laterality	choice	Bilateral | Left | Right | Unilateral | Midline | Not applicable
obs.location	Anatomical location	text	
obs.confidence	Confidence	choice	High | Moderate | Low
obs.notes	Clinical notes / source text	long text	

Clinical examination (repeatable)

Field	Label	Type	Allowed values
exam.type	Examination	choice	General physical | Neurological | Cardiovascular | Respiratory | Abdominal | Musculoskeletal | Dysmorphology | Ophthalmological | Dermatological | Growth assessment
exam.date	Date	date	
exam.examiner	Examiner	text	
exam.source	Evidence source	choice	Clinician-observed | Patient-reported | Caregiver-reported | Documented in a medical record | Derived from a test
exam.findings	Findings	long text	
exam.interp	Clinical interpretation	long text	

Examination measurement (repeatable)

Field	Label	Type	Allowed values
meas.name	Measurement	text	
meas.value	Value	number	
meas.unit	Unit	text	
meas.lo	Reference low	number	
meas.hi	Reference high	number	

Laboratory result (repeatable)

Field	Label	Type	Allowed values
lab.panel	Category	choice	Complete blood count | Biochemical profile | Liver function | Renal function | Electrolytes | Thyroid function | Hormonal | Immunological assay | Metabolic | Amino acid profile | Organic acid profile | Enzyme assay | Cytogenetics | Chromosomal microarray | Karyotype | Molecular diagnostic test
lab.test	Test name	text	
lab.code	Code (e.g. LOINC)	text	
lab.value	Value	text	
lab.unit	Unit	text	
lab.lo	Reference low	number	
lab.hi	Reference high	number	
lab.flag	Abnormal flag	choice	Auto | Normal | Low | High | Abnormal
lab.sample	Sample type	text	
lab.collected	Collected	datetime-local	
lab.lab	Laboratory	text	
lab.method	Method	text	
lab.interp	Interpretation	long text	

Genomic test (repeatable)

Field	Label	Type	Allowed values

test.type	Test	choice	Gene panel | Whole-exome sequencing | Whole-genome sequencing | RNA sequencing | Mitochondrial genome | CNV analysis | Structural variant analysis | Repeat expansion analysis | Methylation profiling
test.lab	Laboratory	text	
test.date	Date	date	
test.status	Status	choice	Planned | Sample received | In analysis | Reported – positive | Reported – negative | Reported – inconclusive
test.assembly	Genome assembly	choice	GRCh38 | GRCh37 | T2T-CHM13
test.panel	Panel code (if panel)	text	
test.result	Result summary	long text	

Data file reference (repeatable)

Field	Label	Type	Allowed values
file.kind	File type	choice	FASTQ | BAM | CRAM | VCF | gVCF | Annotated VCF | CNV / SV file | Interpretation report | Clinical image | Other document
file.uri	Location (URI / path in secure storage)	text	
file.sha256	SHA-256 checksum	text	
file.size	Size	text	
file.consent	Consent covers this file	choice	Yes | No | Unknown
file.note	Note	text	

Genomic variant (repeatable)

Field	Label	Type	Allowed values
variant.gene	Gene symbol	text	
variant.geneId	Gene ID (HGNC / Ensembl)	text	
variant.assembly	Assembly	choice	GRCh38 | GRCh37
variant.chrom	Chromosome	text	
variant.pos	Position	number	
variant.ref	Ref allele	text	
variant.alt	Alt allele	text	

variant.hgvsc	HGVS c.	text	
variant.hgvsp	HGVS p.	text	
variant.zyg	Zygosity	choice	Heterozygous | Homozygous | Hemizygous | Compound heterozygous | Mosaic | Heteroplasmic
variant.ab	Allele balance	number	
variant.consequence	Consequence	text	
variant.inheritance	Inheritance model	choice	Autosomal dominant | Autosomal recessive | X-linked | Mitochondrial | De novo | Unknown
variant.af	Population frequency	text	
variant.afSource	Frequency source	text	
variant.clinvar	ClinVar classification	text	
variant.clinvarReview	ClinVar review status	text	
variant.acmg	ACMG/AMP class	choice	Pathogenic | Likely pathogenic | Uncertain significance | Likely benign | Benign
variant.criteria	ACMG criteria applied	text	
variant.analyst	Analyst	text	
variant.date	Interpretation date	date	
variant.evFor	Supporting evidence	long text	
variant.evAgainst	Conflicting evidence	long text	

Clinical document (repeatable)

Field	Label	Type	Allowed values
doc.title	Title	text	
doc.type	Document type	choice	Clinical note | Discharge summary | Physician report | Referral letter | Patient-reported description | Caregiver-reported description | Laboratory / diagnostic report
doc.date	Date	date	
doc.author	Author	text	
doc.text	Text	long text	

Follow-up visit (repeatable)

Field	Label	Type	Allowed values
follow.date	Date	date	

follow.clinician	Clinician	text	
follow.status	Status	choice	Scheduled | Completed | Missed | Lost to follow-up
follow.summary	Summary	long text	
follow.newFindings	New or changed findings	long text	
follow.plan	Plan	long text	
follow.next	Next follow-up	date	

Consent record (repeatable)

Field	Label	Type	Allowed values
consent.type	Consent for	choice	Clinical genetic testing | Research use | Data sharing (de-identified) | Clinical images | Recontact | Secondary findings
consent.status	Status	choice	Given | Declined | Withdrawn | Pending
consent.date	Date	date	
consent.version	Form version	text	
consent.by	Given by	choice	Patient | Parent / guardian | Legal representative
consent.witness	Witness / recorded by	text	
consent.withdrawn	Withdrawal date	date	
consent.notes	Notes	text	

HPO annotation (attached to an observation)

Field	Meaning
hpo	HPO identifier, e.g. HP:0001250
label	Preferred label at the time of annotation
status	Candidate | Accepted | Rejected | Superseded
conf	Confidence (0–1 for extracted suggestions)
annotator	Person who made or accepted the annotation
date	Annotation date
ver	HPO release used
origin	manual | template | suggested | ai-reviewed | migration | import
replaces	Older identifier this annotation supersedes (migration only)

HPO export columns (TSV / CSV)

Column	Source
PATIENT_ID	Patient ID, or research key in de-identified exports
OBSERVATION_ID	Internal observation identifier
HPO_ID / HPO_TERM	Annotation; empty ID with the original text when unmapped
OBSERVATION_STATUS	Present, Absent, Suspected, Unknown, Not assessed, Not applicable
AGE_OF_ONSET, SEVERITY, FREQUENCY, LATERALITY	Observation fields
EVIDENCE_SOURCE	Who or what the finding comes from
CLINICAL_NOTES, ANNOTATOR	Left empty in de-identified exports
ANNOTATION_DATE, HPO_VERSION	Provenance
REVIEW_STATUS	Accepted, Candidate, Superseded or Unmapped

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