# Ontology and knowledge-resource register

What each external resource is for, its licence position and how release 1.0 uses it.

Licence entries are a working summary. Read the current terms at the source and record the version you load before any production or commercial use. Not every resource is freely reusable.

| Resource | Purpose | Licence (verify at source) | Use in release 1.0 | Source |
|---|---|---|---|---|
| Human Phenotype Ontology (HPO) | Phenotype terms, hierarchy, gene annotations | Free to use with attribution under the HPO licence | Embedded, release 2025-01-16 | hpo.jax.org; new build per release |
| MONDO | Unified disease ontology | CC BY 4.0 | Not embedded; planned for disease coding | mondo.monarchinitiative.org |
| Orphanet / ORDO | Rare-disease nomenclature | CC BY 4.0 | Orphanet counts shown via HPO annotations only | orphadata.com |
| Disease Ontology (DOID) | Disease classification | CC0 | Not used | disease-ontology.org |
| OMIM | Gene–disease catalogue | Licence needed for commercial use and for bulk data | Only OMIM identifiers that HPO distributes are shown; no OMIM text | omim.org |
| SNOMED CT | Clinical terminology | National or affiliate licence | Not embedded | snomed.org |
| ICD-10 / ICD-11 | Diagnosis coding | WHO terms; ICD-11 under CC BY-ND 3.0 IGO | Not embedded; free-text diagnosis fields | who.int |
| LOINC | Laboratory test codes | Free under the LOINC licence | Code field provided; table not embedded | loinc.org |
| RxNorm | Medication names | Free; some sources need a UMLS licence | Not embedded | nlm.nih.gov |

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| Uberon / FMA / Cell Ontology / GO | Anatomy, cells, gene function | Open (CC BY) | Not used in release 1.0 | obofoundry.org |
| ClinVar | Variant clinical significance | Public domain (NCBI) | Fields for classification and review status; no live lookup | ncbi.nlm.nih.gov/clinvar |
| ClinGen / GenCC | Gene–disease validity | CC0 | Not embedded; recommended next for panel curation | clinicalgenome.org; thegencc.org |
| DECIPHER | Genotype–phenotype evidence | Access agreement; commercial use needs a licence | Not used | deciphergenomics.org |
| Genomics England PanelApp | Gene panels | Terms restrict commercial use | Not used; Tribro-Omics panels are curated independently | panelapp.genomicsengland.co.uk |
| Exomiser | Phenotype-driven variant ranking | AGPL-3.0 | Reached by Phenopacket export; not bundled | exomiser.github.io/Exomiser |
| LIRICAL | Likelihood-ratio phenotype analysis | Open source; check licence text | Reached by Phenopacket export | github.com/TheJacksonLaboratory/LIRICAL |
| Phen2Gene | Phenotype-to-gene ranking | MIT | Not bundled; HPO list export is compatible | github.com/WGLab/Phen2Gene |
| Monarch Initiative | Integrated phenotype knowledge | Open; source-specific terms apply | Not used | monarchinitiative.org |

## Rules applied

- Every annotation stores the ontology release it was made with.
- A new release never rewrites old annotations; a migration report is reviewed first.
- Attribution: “This product uses the Human Phenotype Ontology (hpo.jax.org)” appears in the tool, the reports and the exports.
- For each resource added later, record: authoritative source, version, licence, update frequency, download or API method, attribution text and permitted use.

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