

SN	Table names	Columns/Attributes	Indexes/Unique	Description
1.	cell_type_enrichment_level1	<ul style="list-style-type: none"> <li>• level1_id (PK) BIGINT</li> <li>• cell_type_name VARCHAR(128) NOT NULL</li> <li>• lineage VARCHAR (8) NOT NULL</li> <li>• enrichment_score DECIMAL(6,4)</li> </ul>	<ul style="list-style-type: none"> <li>• PK: level1_id</li> <li>• UNIQUE: (cell_type_name)</li> </ul>	
2.	Cell_type_enrichment_level2	<ul style="list-style-type: none"> <li>• level2_id (PK) BIGINT</li> <li>• level1_id (FK) BIGINT NOT NULL</li> <li>• cell_type_name VARCHAR(128) NOT NULL</li> <li>• enrichment_score DECIMAL(6,4)</li> </ul>	PK: level2_id UNIQUE: (level1_id, cell_type_name)	
2.	disease	<ul style="list-style-type: none"> <li>• disease_id (PK) BIGINT</li> <li>• disease_name VARCHAR (128) NULL</li> <li>• category VARCHAR (32) NULL (e.g. “autoimmune”, “CH”, etc)</li> </ul>	<ul style="list-style-type: none"> <li>• PK: disease_id</li> <li>• UNIQUE: (disease_name)</li> </ul>	Catalogue of autoimmune / autoinflammatory diseases and related diagnostic labels. Provides a controlled list of disease names and broad categories (e.g. “autoimmune”, “autoinflammatory”) used to annotate patients, studies, and variant–disease associations.
3.	genes	<ul style="list-style-type: none"> <li>• gene_id (PK) BIGINT</li> <li>• ref_genome_id, FK, NOT NULL</li> <li>• gene_symbol VARCHAR (32) NOT NULL</li> </ul>	<ul style="list-style-type: none"> <li>• PK: gene_id</li> <li>• UNIQUE: (gene_symbol, ref_genome_id) – same symbol can exist on</li> </ul>	Registry of genes (one row per gene symbol) that variants and annotations reference. Ensures standardized gene naming across studies and annotation sources (HGNC-style symbols like STAT3, DNMT3A, TET2).

			<p>different genome builds</p> <ul style="list-style-type: none"> <li>• INDEX: idx_genes_ref_genome(ref_genome_id)</li> </ul>	
4.	sequencing_experiment	<ul style="list-style-type: none"> <li>• seq_exp_id (PK) BIGINT</li> <li>• sample_id FK NOT NULL</li> <li>• study_id (FK) BIGINT</li> <li>• technology ENUM('GWAS','Tapestri','GT_seq','WES','WGS','Other') NOT NULL</li> <li>• coverage DECIMAL</li> </ul>	<ul style="list-style-type: none"> <li>• PK: seq_exp_id</li> <li>• INDEX: idx_seqexp_sample(sample_id)</li> <li>• INDEX: idx_seqexp_study(study_id)</li> <li>• INDEX: idx_seqexp_technology(technology)</li> </ul>	Each sequencing run / library prepared from a sample under a given study (paper/cohort). This is the anchor for downstream per-experiment cell types, variant calls, clonality and summaries.
5.	sample_variant_call	<ul style="list-style-type: none"> <li>• call_id (PK) BIGINT</li> <li>• cell_type_id (FK) BIGINT NOT NULL</li> <li>• seq_exp_id (FK) BIGINT NOT NULL</li> <li>• level1_id BIGINT</li> <li>• level2_id BIGINT</li> <li>• variant_id (FK) BIGINT NOT NULL</li> <li>• vaf DECIMAL(4,3) NULL</li> <li>• variant_origin VARCHAR(32) NULL</li> </ul>	<ul style="list-style-type: none"> <li>• PK: call_id</li> <li>• UNIQUE: (seq_exp_id, variant_id, cell_type_id) – one call per variant/celltype/experiment</li> <li>• INDEX: idx_call_cell_type(cell_type_id)</li> <li>• INDEX: idx_call_variant(variant_id)</li> </ul>	Stores individual variant calls observed in a specific sequencing experiment, within a specific cell-type compartment. Each row links a variant to an experiment and cell type, with an associated VAF and call-level origin (somatic/germline). The unique constraint ensures you don't duplicate the same variant-cell-type call within the same experiment.

7.	study	<ul style="list-style-type: none"> <li>study_id BIGINT (PK)</li> <li>study_name VARCHAR (128) name</li> <li>pmid VARCHAR(16) NULL</li> <li>year INT NULL</li> <li>notes TEXT NULL</li> </ul>	<ul style="list-style-type: none"> <li>PK: study_id</li> <li>UNIQUE: (pmid)</li> <li>INDEX: idx_study_year(year)</li> </ul>	Projects / publications / cohorts under which experiments are organized. Stores a human-readable name, PubMed ID, year and any notes so that sequencing experiments, patients and downstream analyses can be grouped and reported at the study level.
9.	variant_annotation	<ul style="list-style-type: none"> <li>ann_id (PK) BIGINT NOT NULL</li> <li>variant_id (FK) BIGINT</li> <li>hgvs VARCHAR (255) NOT NULL</li> <li>consequence VARCHAR (32) (e.g., missense_variant, splice_donor_variant) NOT NULL</li> </ul>	<ul style="list-style-type: none"> <li>PK: ann_id</li> <li>INDEX: idx_varann_variant(variant_id)</li> </ul>	Stores per-variant functional annotations: gene assignment, HGVS protein change and predicted consequence, plus optional mapping to a protein domain. The UNIQUE (variant_id) enforces a single “canonical” annotation row per variant (e.g. one main gene / consequence / protein HGVS per allele).
10.	variants	<ul style="list-style-type: none"> <li>variant_id [PK] BIGINT NOT NULL AUTO_INCREMENT</li> <li>protein_domain_id FK BIGINT NOT NULL</li> <li>gene_id FK BIGINT NOT NULL</li> <li>ref_genome_id [FK] INT NOT NULL</li> <li>chrom VARCHAR (8) NOT NULL</li> <li>pos INT NOT NULL</li> </ul>	<ul style="list-style-type: none"> <li>PK: variant_id</li> <li>UNIQUE: (ref_genome_id, chrom, pos, ref, alt) – ensures no duplicate variants per genome build</li> <li>INDEX: idx_variant_chrom_pos (chrom, pos)</li> </ul>	Central hub of genomic variants: one row per allele defined by genome build, chrom, position, ref and alt, with links to a reference-genome table and a variant-type table (SNV, indel, etc.). All downstream objects (calls, annotations, disease associations, functional validation) reference this table.

		<ul style="list-style-type: none"> <li>ref VARCHAR (255) NOT NULL</li> <li>alt VARCHAR (255) NOT NULL</li> <li>variant_origin VARCHAR(32) NOT NULL</li> <li>vaf DECIMAL(4,3)</li> </ul>		
11.	protein_domain	<ul style="list-style-type: none"> <li>protein_domain_id PK BIGINT</li> <li>domain_name VARCHAR(16) NULL</li> <li>domain_type VARCHAR(25) NULL</li> </ul>	<ul style="list-style-type: none"> <li>PK: protein_domain_id</li> <li>UNIQUE: (domain_name)</li> </ul>	Lookup table for protein domains affected by variants, such as SH2 domains or Pfam/InterPro regions. Enables annotation of which structural/functional region of the protein is altered by a given variant (via variant_annotation.protein_domain_id).
12.	sample	<ul style="list-style-type: none"> <li>sample_id PK BIGINT</li> <li>disease_id FK BIGINT NOT NULL</li> <li>patient_code VARCHAR(8) NULL</li> <li>tissue_source VARCHAR(16) NOT NULL</li> <li>sample_type VARCHAR(64) NULL</li> </ul>	<ul style="list-style-type: none"> <li>PK: sample_id</li> <li>INDEX: idx_sample_patient(patient_id)</li> </ul>	Biological samples collected from patients (e.g. PBMC, bone marrow, intestinal biopsy). Each sample belongs to one patient and can be sequenced multiple times. sample_type differentiates bulk DNA, single-cell DNA, TCR sequencing, etc.

14.	Functional_validation	<ul style="list-style-type: none"> <li>• validation_id PK BIGINT</li> <li>• variant_id FK BIGINT NOT NULL</li> <li>• assay_type VARCHAR(64) NULL</li> <li>• result VARCHAR(32) NULL</li> </ul>	<ul style="list-style-type: none"> <li>• PK: validation_id</li> <li>• INDEX: idx_fv_variant(variant_id)</li> </ul>	Captures experimental evidence from functional assays for specific variants (e.g. reporter assays, phosphorylation readouts, CRISPR screens). Multiple assays can exist per variant, with the result summarised as gain-of-function, loss-of-function, neutral, etc.
15.	reference_genome	<ul style="list-style-type: none"> <li>• ref_genome_id BIGINT PK, auto-inc, NOT NULL</li> <li>• ref_genome_name VARCHAR(32) NOT NULL</li> <li>• source VARCHAR(64) NULL</li> <li>• version VARCHAR(16) NULL</li> <li>• notes TEXT NULL</li> </ul>	<ul style="list-style-type: none"> <li>• PK: ref_genome_id</li> <li>• UNIQUE: (ref_genome_name)</li> </ul>	