SwissGenVar catalogs and data sources

Whenever applicable, SwissGenVar uses the existing international and/or SPHN standards ("Pre-existing catalogs and data sources"). When there was no satisfying data standard available, the SwissGenVar consortium has adapted or created those relevant to the project ("Internal catalogs").

Pre-existing catalogs and data sources

Information	Data source	Obtained from	Full name	Description
Causality	OMIM	morbidmap.txt	Online Mendelian Inheritance in Man	The genes causality is retrieved from OMIM
Clinical indication	HPO		Human Phenotype Ontology	Standardized vocabulary of phenotypic abnormalities encountered in human disease
Clinical significance	ClinVar	VEP		The ClinVar pathogenicity, using the 5 ACMG levels, is displayed
Diagnosis	OMIM		Online Mendelian Inheritance in Man	
Ethnicity	GnomAD			African/African-American, Amish, Latino/Admixed American, Ashkenazi Jewish, East Asian, Finnish, Non-Finnish European, Middle Eastern, South Asian, Other
Frequency	GnomAD	VEP		The minor allele frequency comes from GnomAD
Gene name	HUGO	VEP	HUman Genome Organisation	Genes are uniquely named according to the HUGO Gene Nomenclature
LRG sequence	LRG	EBI REST API (id)	Locus Reference Genomic	A Locus Reference Genomic (LRG) record contains stable reference sequences that are used for reporting sequence variants with clinical implications. Documentation available on https://www.lrg-sequence.org/web-service/ and terms of use are readable on https://www.ebi.ac.uk/about/terms-of-use/ Example request : https://www.ebi.ac.uk/ebisearch/ws/rest/lrg?query=NM_004972.3&format=json
Phenotype	HPO	HPO API ()	Human Phenotype Ontology	Standardized vocabulary of phenotypic abnormalities encountered in human disease
Transcripts	RefSeq, Ensembl	VEP		Transcripts from both Refseq and Ensembl are referenced
Variant description	HGVS	VEP	Human Genome Variation Society	This nomenclature is used for the description of sequence variants (namely HGVSg., HGVSc. and HGVSp.). It will be complemented by ICSN (International System for Human Cytogenetic Nomenclature) for large genomic aberrations

Internal catalogs

Information	Possible values	Description
Aneuploidies	Yes; No	
Canton	List of Swiss cantons, plus "Non-Swiss"	
Chromosomal sex	XX; XY; Other	
Clinical sex	Male, Female, Ambiguous	
Karyotypic sex	45X, 46XX, 46XY, 47XXY, 47XYY, 47XXX	This field is an expandable list, and its content is conditional to the value of "Chromosomal sex"
Clinical status	Affected; Partially affected; Not affected	
Co-occurences	Yes; No	
Collection method	Case-control; Clinical testing; Reference population; Research; Other; Unknown	
Consent	Yes; No	
Detection method	Sequencing; Fragment analysis; Southern Blot; Conventional cytogenetics; FISH (IFISH or MFISH); Array (Oligo or SNP); qRTPCR; MLPA; NGS-based CNV detection (Panel/WES/WGS); Other; Not performed	
Gene locus type	Protein coding gene; Non-coding RNA gene; Long non-coding RNA; MicroRNA; Ribosomal RNA; Transfer RNA; Small nuclear RNA; Small nucleolar RNA: Other; Locus subjected to imprinting	Partly coming from HGNC
Index patient	Yes; No	
Inheritace of the disease	AD - Autosomal dominant, AR - Autosomal recessive, PD - Pseudoautosomal dominant, PR - Pseudoautosomal recessive, DD - Digenic dominant, DR - Digenic recessive, IC - Isolated cases, ICB - Inherited chromosomal imbalance, Mi - Mitochondrial, Mu - Multifactorial, SMo - Somatic mosaicism, SMu - Somatic mutation, XL - X-linked, XLD - X-linked dominant, XLR - X-linked recessive, YL - Y-linked	From OMIM
Inheritance of the variant	De novo constitutive; De novo mosaic; Paternally inherited, constitutive in father; Paternally	

	inherited, mosaic in father; Maternally inherited, constitutive in mother; Maternally inherited, mosaic in mother; Biparental; Imbalance arising from a balanced parental rearrangement; Inherited mosaic; Unknown	
Locus subjected to imprinting	Yes; No; Unknown	
Submitting institution	One acronym per partner institution	
Variant effect	Missense variant; Nonsense variant; Stop loss; Start loss; Intron variant; Frameshift variant; Inframe insertion; Inframe deletion; Splicing variant; Synonymous variant; Regulatory variant; Non-coding transcript variant; Extension: Repeat expansion	Not compulsory; Intron variant stands for not splicing variants only, the rule to define splicing variants will have to be defined
Variant location	Upstream gene; Downstream gene; 5 prime UTR; 3 prime UTR; Promoter region; Intronic region; Coding region; Splicing region; Regulatory region; Intergenic region	Not compulsory
Variant type	Substitution; Deletion; Insertion; Duplication; Inversion; Conversion; Deletion-insertion; Repeat variation; Complex; Methylation/epigenetic change; CNV-deletion; CNV-duplication; CNV-amplfication; CNV-complex rearrangement; CNV-insertion; CNV-insertion/duplication; SV-translocation; SV-inversion; Aneuploidy	Adapted from HGVS
Variant zygosity	Heterozygous; Homozygous; Hemizygous; Mitochondrial heteroplasmy; Mitochondrial homoplasmy; Unknown; Mosaic; Chimeric; Ambiguous	