

The upgrade you've been waiting for.

SeqOne Platform's germline analysis pipeline, GermVar, builds on everything Congenica Rare Disease (CRD) offers today, with a significantly enhanced feature set. With multi-variant-type calling, AI-powered prioritisation, flexible filtering, automated re-analysis, and up-to-date integrated tools through consistent release cadence, GermVar is designed to help your teams work more efficiently and reach diagnoses with greater confidence.

- ✓ CE-IVD certified under IVDR (Class C)
- ✓ Your data stays yours, with a dedicated, single-tenant instance available
- ✓ 100% of CRD capacities preserved or enhanced
- ✓ Broad vendor compatibility
- ✓ Explainable AI prioritisation & automated ACMG classification
- ✓ 20+ new capabilities available from day one

Read on for a detailed, side-by-side comparison to help you evaluate the transition with confidence.

Secondary Analysis

Variant Type	CRD	GermVar	Comparison
SNV/Indel	Sentieon DNaseq (Panels & WES)	Sentieon DNAScope + ML (Panels, WES & WGS)	Enhanced
CNV	ExomeDepth (SaaS only)	Adapted callers per data type	Enhanced
Structural Variants	Not available	Delly + Cue	GermVar exclusive
STR	Not available	STRchive v2.16.0 (74 loci)	GermVar exclusive
SegDup genes	Not available	Sentieon caller (9 genes: SMN1, PMS2, CYP2D6, GBA, STRC...)	GermVar exclusive
Mitochondrial	Not available	Mutect2 pipeline	GermVar exclusive
MEI	Not available	AluMEI	GermVar exclusive
Mosaic	Not available	TNScope-based (3,800+ genes)	GermVar exclusive

Note: GermVar is one unified pipeline for Panels, WES & WGS plus UMI processing.

Tertiary Analysis (VCF Import)

Feature	CRD	GermVar	Comparison
SNV/Indel	Yes (v4.1+)	Yes	Equal
CNV	Yes	Yes	Equal
Structural variants	Can load	Can load	Equal
STR/Repeats	Can load	Can load	Equal
Long-read VCF	Not supported	ONT & PacBio	GermVar exclusive
Phased variants	Not supported	Loaded & visualized	GermVar exclusive
Splicing predictions	5 algorithms	Dedicated viewer	Equal

Note: GermVar adds Dragen compatibility and WES capture-kit selection for VCF imports.

AI-Powered Prioritisation

Feature	CRD	GermVar	Comparison
Pathogenicity Scoring	Congenica AI + Exomiser (confidence rating, contribution scoring)	UP ² pathogenicity scoring (-1 to 1), retrained on 3M+ variants	Enhanced
Gene-Phenotype Matching	Exomiser	Phenogenius v3.1: 6,181 genes, 19,409 HPO terms	Enhanced
Non-coding Variants	Heuristic ACMG hints (top 100 variants)	Non-coding via GPN-MSA	Enhanced
HPO Extraction	N/A	Automatic HPO extraction from text	GermVar exclusive
Explainability	Limited	Explainable AI	GermVar exclusive
Performance Benchmark	57% of causal variants in top 10 (Genomics England WGS)	98% of causal variants in top 10 (Genomics England WGS)	Enhanced
Benchmark Comparison (DiagAI vs Exomiser, x565 Genomics England WGS singletons)	81% ranked #1	98% in Top-10, +42% relative improvement at Top-1 download the white paper	Enhanced

AutoACMG (Automated Classification)

Feature	CRD	GermVar	Comparison
Criteria Scoring	Heuristic Exomiser hints, top 100 variants only, partial criteria	All 28 ACMG/AMP criteria scored on every variant	Enhanced
Classification	No pre-computed 5-tier class	Pre-computed 5-tier class	GermVar exclusive
Justification	No per-criterion justification	Per-criterion justification	GermVar exclusive
Standards	No ClinGen VCEP	ClinGen VCEP rules	GermVar exclusive
Validation	Manual	One-click validate/override	GermVar exclusive

Family & Trio Analysis

Feature	CRD	GermVar	Comparison
Trio Analysis	DeNovoGear	GermlineFamily <i>up to 7 family members for the same price</i>	Enhanced
De Novo Detection	Automated	Automated	Equal
Inheritance Analysis	Yes	Yes	Equal
Compound Het	Yes	Grouped (small+large) with VAF, coverage & quality	Enhanced
Genotype & Inheritance Display	Yes	Trio colours, sex-chromosome inheritance, Mendelian-violation warnings, hemizygous labels	Enhanced
Segregation	Segregation tab	Yes	Equal
Family Verification	Yes	Relationship coefficient	Equal

Annotation & Databases

Database	CRD	GermVar	Comparison
gnomAD	v2.1.1 exome	v4.1 (exome+genome)	Enhanced
ClinVar	Static release	2026-02 (quarterly)	Enhanced
OMIM	Mar 2023	2026-03	Enhanced
PanelApp	Not integrated	England + Australia (2026-03)	GermVar exclusive
AlphaMissense	Not available	Yes	GermVar exclusive
UP ²	Not available	Yes	GermVar exclusive
REVEL/CADD/SIFT/PolyPhen	Yes	Yes	Equal
SpliceAI	Yes	CI-SpliceAI	Enhanced
Constraint (LOEUF/Z-missense)	Yes	gnomAD v4.1 (LOEUF slider, Intolerant/Tolerant badges)	Enhanced
Internal lab frequency	View only	Filterable	Enhanced
CNV ACMG classification	Not available	Yes	GermVar exclusive
gnomAD SV	Not available	Yes	GermVar exclusive

Note: all databases update quarterly, independent of platform releases.

Filtering & Presets

Filter Type	CRD	GermVar	Comparison
Gene & Region	3 filters	5 filters	Enhanced
Population Frequency	12 filters	5 filters	Equal
Prediction Scores	11 filters	9 filters	Equal
Quality Filters	3 filters	5 (adds allelic ratio, GQ, strand bias)	Enhanced
Classification & AI	8 filters	9 (AutoACMG + Variant Knowledge Base)	Enhanced
Presets & Management	5 filters	7 (combinable profiles, 11 built-in presets, favourites, preset locking, public/private sharing)	Enhanced

Quality Control

QC Metric	CRD	GermVar	Comparison
Coverage Analysis	Configurable depths	Gene-level (avg/worst-exon), gene/exon XLS exports, WES summaries in reports	Enhanced
Contamination Check	No	Built-in	GermVar exclusive
Sample Identity (ID-Check)	No	Genetic markers	GermVar exclusive
Configurable QC Thresholds	No	Per project	GermVar exclusive

Workflow & Reporting

Feature	CRD	GermVar	Comparison
Double-user check	Yes	Yes	Equal
Analysis assignment	No	Yes	GermVar exclusive
Analysis locking	No	Yes	GermVar exclusive
Protocol module	No	Yes	GermVar exclusive
Clinical PDF report	Yes	Yes	Equal
Technical report	No	Yes	GermVar exclusive
ID-Check report	No	Yes	GermVar exclusive
Custom report sections	No	Yes	GermVar exclusive
Public API	REST v3	Public API v1 (12 endpoint categories + API keys)	Equal
Define User Access Levels	Yes	Yes	Equal

Your Wishlist, Delivered

- ✓ Combinable filter profiles
- ✓ Automated re-analysis of negative cases (GenomeAlert)
- ✓ CNV calling for whole genomes
- ✓ Mitochondrial variant calling
- ✓ Internal lab-frequency filtering
- ✓ Cross-sample variant search
- ✓ HPO-based variant filtering
- ✓ Sample identity & kinship check (ID-Check)
- ✓ ACMG classification for CNVs
- ✓ SV population-frequency filtering
- ✓ Long-read sequencing support
- ✓ Flexible multi-VCF upload

And many more, at each release.

Ready for the next step?

Contact us

SeqOne Platform is a software-based in vitro diagnostic medical device, CE-marked as a Class C under the Regulation (EU) 2017/746, designed to support healthcare professionals in analyzing and interpreting raw genomic and sequencing data from patients undergoing NGS or CGH array analysis. By providing qualitative and actionable insights, the platform functions as a decision support system, aiding healthcare professionals in making clinical decisions. GermVar is a feature of SeqOne Platform. Find out more about our compliance and security framework at trust.seqone.com. Manufacturer: SeqOne (France)