

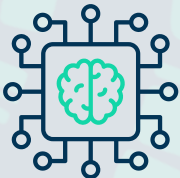
## FOR RARE & INHERITED DISEASES



Web-based  
User-friendly interface



One-stop shop  
germline and somatic



Powered by  
explainable AI

SeqOne

Project name > GermVarTrio

QC GeneCov Variants CNVs Clinical Reports Working files

Filters 4 Variants 43/271 141 Genes 12/23

	DIAG.AI	ACMG	VKB	PATIENT GT VAF	GENE	TRANSCRIPT	HGVs	EXON	EFFECT	CLINVAR
	96	5	5	50.8%	SLC25A22	NM_024698.6	c.813_814del p.A272QfsTer144	9/10	High ● Frameshift	3 LP Mucopolysaccharidosis VI
	90	4	5	46.9%	SLC25A22	NM_024698.6	c.818G>A p.R273K	9/10	Moderate ● Missense	
	85	5	5	100%	BCAP31	NM_001139441.1	c.-45+1_-45+2d -	N/A	High ● Splice Donor	? VUS
	75	5	5	0%	OCLN	NM_002538.4	c.1037+1G>A -	N/A	High ● Stop Gained	
	71	4	4	44.8%	SLC3A1	NM_000341.4	c.1084C>T p.R396W	6/10	Moderate ● Stop Gained	Primarily of unknown significance
	71	3	4	88.2%	MEFV	NM_000243.3	c.169_17@2del p.A744S	3/5	High ● Frameshift	
	70	5	5	43.8%	METTL23	NM_001080510.5	c.413A>G p.G303R	3/3	High ● Stop Gained	
	52	4	5	56%	TLR7	NM_004333.6	c.907G>A p.R396W	5/12	Moderate ● Stop Gained	3 LP Mucopolysaccharidosis VI
	51	5	New	44.6%	DHCR7	NM_004333.6	c.2102G>A p.R86Ter	8/9	High ● Stop Gained	
	50	4	New	50%	CYP24A1	NM_016562.4	c.2144-10delinsTT -	9/12	High ● Frameshift	2 B 1
	50	5	New	54%	CEP55	NM_000552.5	c.991C>T p.P301LfsTer139	9/9	Moderate ● Stop Gained	8 P 1 Primarily of unknown significance
	23	3	New	50.6%	RAB3GAP1	NM_004333.6	c.163del p.R1046C	1/16	Moderate ● Frameshift	
	22	3	New	52.7%	VWF	NM_152641.4	c.5237G>A p.P703S	5/12	Moderate ● Stop Gained	
	14	3	New	51.5%	JPH3	NM_004333.6	c.2107C>T p.R1746K	4/9	Low ● Stop Gained	5 P 1 Congenital hypothyroidism
	11	3	New	51.5%	JPH3	NM_004333.6	c.2107C>T p.R1746K	4/9	Low ● Stop Gained	5 P 1 Congenital hypothyroidism
	11	3	New	51.5%	OTOF1	NM_173591.7	c.5237G>A	4/9	High	3 LP Mucopolysaccharidosis VI

# WHO WE ARE



## OUR VISION

Founded in 2017, SeqOne develops state-of-the-art genomics analysis tools for clinical applications in oncology and rare diseases. **Our vision is to make personalized healthcare a reality** for all by democratizing access to **advanced genomic insights**, enabling every lab to offer **affordable** precision medicine solutions to patients **at scale**.

## OUR SOLUTION

Our **CE-IVD** platform provides **time-saving, flexible, and cost-effective** decision support, **empowering decentralized laboratories** worldwide to deliver precision diagnostics. We integrate **cutting-edge AI and big data technologies** to redefine genetic data analysis.

70+

SeqOne employees



1000+ routine users



Fast-growing, present in 10 countries

# THE SEQONE PLATFORM

Complete germline pipelines for **DNA** analysis, tailored for **targeted panels, exome, genome and aCGH arrays** to provide support for diagnosis and therapeutic decisions.

Hereditary Cancers

Rare & Inherited Diseases

GermVar  
(from FASTQ file)

In-house / Third-party secondary analysis

GermVar Tertiary  
(VCF from Dragen, Epi2ME, Alissa Reporter or XLS file)

# ALL-IN-ONE NGS DATA ANALYSIS SOLUTION

## For Solo & Family Analyses



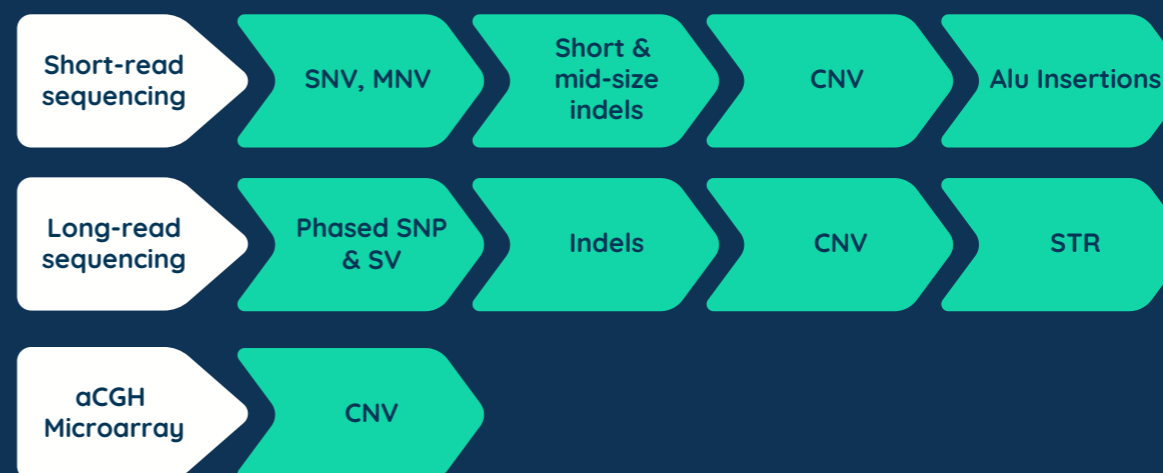
### Maximize your workflow efficiency

- **SeqOne SDS** automates data and metadata upload from the sequencer, autorun analysis, and bi-directional communication with the LIMS system
- **DiagAI machine learning assistance** to rank, shortlist and suggest causal variants
- **Advanced & customizable filtering and reporting** to fit with lab workflow
- **Private and secure Variant Knowledge Base (VKB)** to keep track of past evaluations



### Ensure accurate identification of your variants

- **Automated classification** based on ACMG/ClinGen
- **Premium annotation data sources:** ClinVar, gnomAD V4, OMIM
- **Scoring integration:** REVEL, CI-SpliceAI
- **Pathogenicity prediction** with an in-house, AI-powered score
- **Precise variant detection** on Hg19/Hg38 reference genome



### Benefit from a flexible and future-proof platform

- **Wet-lab agnostic** (Illumina, Agilent, Twist, Roche, etc.)
- **Sequencer compatibility** (Illumina, MGI, Element, ONT, etc.)
- **Monthly updates** to keep pace with scientific discoveries

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## CONFIDENTLY PERFORM NGS DATA ANALYSIS

### Regulatory Compliance

CE-IVD platform, ISO-13485 certified

### Cyber Security

ISO-27001 certified hosting environment  
Health Data Storage (HDS) certified  
Patented Genomic Data Double Encryption

### Data Privacy

Data not shared or commercialized  
Variant knowledge base remains fully private

## SEQONE genomeAlert!



Automated variant reevaluation agent developed in collaboration with geneticists



Monthly reanalysis of all historical cases (ClinVar latest updates)



Time-saving report (highlight new candidates or revoke diagnoses)