

CENTOGENE
THE RARE DISEASE COMPANY



Bioinformatic SaaS Platform

CentoCloud[®]

The Decentralized Diagnostic Solution



Diagnose Inherited Diseases, No Matter Where You Are Located

Analyze, interpret, and report genomic variants via our decentralized Software as a Service (SaaS) platform. With CentoCloud, we enable laboratories around the world to establish and run state-of-the-art genomic testing and deliver the best diagnostic insights to local patients. As a cloud-based SaaS platform, CentoCloud is specifically designed to support laboratories with the execution of superior Next Generation Sequencing (NGS)-based diagnostics. Our fully automated bioinformatics pipeline analyzes NGS data based on CENTOGENE's validated testing products, annotated with data from the CENTOGENE Biodatabank.

CentoCloud Technical Specifications

Below you will find details regarding the Illumina sequencing platform and available library kits that can be used in combination with CentoCloud.

			NGS Panel	WES	WGS	
Technology Platform	Illumina	MiSeq	✓			
		NextSeq	✓	✓	✓	
		HiSeq	✓	✓	✓	
		NovaSeq		✓	✓	
Library Kits	Illumina	DNA Prep with Exome 2.0 Plus Enrichment		✓		
		DNA Prep with Exome 2.0 Plus Enrichment plus Mitochondrial Panel		✓		
		Nextera Rapid Capture Exome v1.2		✓		
		DNA Prep, DNA PCR-Free Prep or TruSeq DNA PCR-Free			✓	
	Agilent	SureSelect Human All Exon V5, V6, V7, V8		✓		
	IDT	xGen Exome Research Panel v2		✓		
	Twist Bioscience	Twist Alliance CNTG Exome			✓	
		Twist Alliance CNTG Rare Disease Panel	✓			
		Twist Alliance CNTG Hereditary Oncology Panel	✓			
		Human Core Exome		✓		
		Human Comprehensive Exome		✓		
		Exome 2.0		✓		

Required Input

- Details on sequencing platform and library kit used for sequencing the sample
- Patient details along with a description of the patient phenotype using Human Phenotype Ontology (HPO) terms
- 2 FASTQ files per patient sample
- Patient consent for analysis to be performed by CENTOGENE
- For CNV calling, for whole exomes from Illumina, Agilent, IDT, and Twist Exome 2.0 library kits, to ensure the highest quality reporting of such variants, we require 50 reference samples before CNVs can be reported

Key Service Deliverables

As part of CENTOGENE's promise to best-in-class diagnostics, medical reporting with clear diagnostic statements are provided by CENTOGENE's team of expert medical geneticists. For every diagnostic test, you will be provided with the processed data files generated by our bioinformatics pipeline. Sample output files from CentoCloud based on analysis of Genome in a Bottle Consortium (GIAB) data can be requested by contacting your CENTOGENE sales representative.

For CentoCloud Exome, Rare Disease Panel, and Hereditary Oncology Panel

File Extension	Description	No. Expected Files
.bam	Alignments containing nucleotide sequences with position-specific quality scores as well as genomic location	1
.bai	Indexing file accompanying any .bam file, used for quickly accessing nucleotide sequences in a given genomic location	1
.pass.vcf	Corresponds to the quality-checked vcf files from GATK's haplotype caller (hpl) or Freebayes (freb)	2
*.xls	<ul style="list-style-type: none">• Tabular-separated text file containing basic sequencing statistics, such as "total number of reads", "number of mapped reads", etc.• Gender and kinship check results• List of variants with various annotations, incl. genotype, zygosity, quality scores, VAF, dbSNP id, Gnomad HPO Match count, pre-classification, clinical prioritisation, etc.	4
*.txt	Coverage metrics (mean, 10x, 20x)	1

For CentoCloud Genome

File Extension	Description	No. Expected Files
.bam	Alignments containing nucleotide sequences with position-specific quality scores as well as genomic location	1
.bai	Indexing file accompanying any .bam file, used for quickly accessing nucleotide sequences in a given genomic location	1
.pass.vcf	Corresponds to the quality-checked vcf files from Dragen	1
*.xls	<ul style="list-style-type: none">• Tabular-separated text file containing basic sequencing statistics, such as "total number of reads", "number of mapped reads", etc.• Gender and kinship check results• List of variants with various annotations, incl. genotype, zygosity, quality scores, VAF, dbSNP id, Gnomad HPO Match count, pre-classification, clinical prioritisation, etc.	4
*.txt	Coverage metrics (mean, 10x, 20x)	1

For More Information

centogene.com

For Ordering

centoportal.com

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