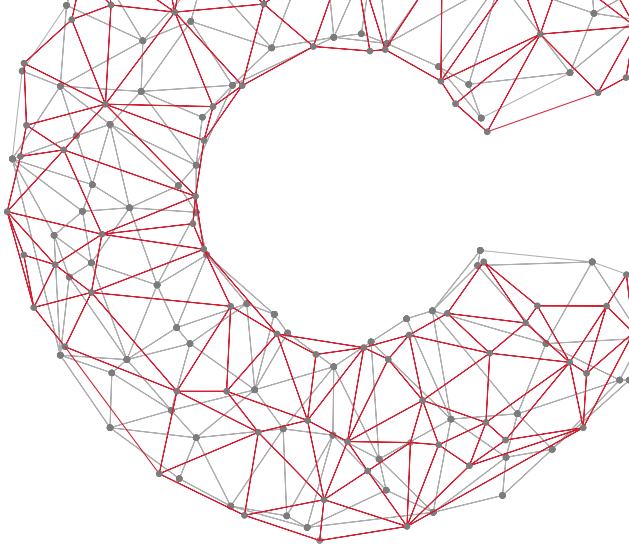
CENTOGENE
THE RARE DISEASE COMPANY



CentoCloud

The Decentralized Diagnostic Solution

TECHNICAL SHEET

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	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	CENTOGENE Exome (CentoXome)		✓	
		CENTOGENE Rare Disease Panel	✓		
		CENTOGENE Hereditary Oncology Panel	✓		

Required input:

- Details on sequencing platform and library kit used for sequencing the sample
- Patient details along with clinical information (at least 4 – 5 HPO terms describing the patient phenotype)
- 2 FASTQ files per patient sample
- Patient consent for analysis to be performed by CENTOGENE
- For CNV calling, 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 (hpcl) or Freebayes (freb)	2
*.xls	• 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.	3
*.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
.vcf	Corresponds to the quality-checked vcf files from Dragen	1
*.xls	• 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.	3
*.txt	Coverage metrics (mean, 10x, 20x)	1