NGS Panels

A Targeted Approach For Testing Genetic Disorders

PRODUCT SHEET

NGS Panels
Next Generation Sequencing

CENTOGENE's Next Generation Sequencing (NGS) gene panels are designed to reflect the fast-growing knowledge of complex associations of genes with diseases as well as maximize clinical sensitivity.

Our panels have been optimized to test for a wide selection of hereditary genetic disorders across 16 different disease categories and follow a phenotype-directed approach that includes all relevant clinical genes. Additionally, we have included genes necessary for differential diagnosis of syndromes with overlapping phenotype(s) – enabling the diagnosis of a disease that could have otherwise been missed. Our high-quality sequencing is supported by complementary assays to provide advanced detection and enable a true diagnosis. This approach maximizes the clinical utility, de-risks panel choice, increases cost-effectiveness, and ultimately simplifies the diagnostic process.

The CENTOGENE Advantage

- Coverage of **all relevant disease-causing genes** and non-coding and coding pathogenic variants
- The most **up-to-date panel gene content** with the latest medical and in-house findings
- **High-quality analysis for precise clinical interpretation** using advanced bioinformatics and artificial intelligence-powered tools
- **Best-in-class insights** powered by the world's largest rare disease-centric Bio/Databank from the leader and trusted partner in rare disease diagnostics
- **Dedicated team of rare disease experts** to provide the best clinical interpretation and life-long support

NGS-Panel Options


Cardiovascular


Dermathology


Dysmorphology


Ear, Nose & Throat


Endocrinology


Hematology


Hepatology


Immunology


Metabolic Disorders


Nephrology


Neurology


Oncology


Ophthalmology


Osteology


Pneumology


Reproductive Genetics

Key Features and Performance

COVERAGE	• ≥ 99.5% targeted regions covered at ≥ 20x • Mean depth coverage 150x – 1000x • For each gene, all SNVs described in HGMD® and our rare disease-centric Bio/Databank are covered, including relevant deep intronic and regulatory variants.
GENES	For a complete overview of included genes, please visit: www.centogene.com/ngspanels-medical-reporting
SPECIFICITY	≥ 99.9% guaranteed for all reported variants. Variants with low quality and/or unclear zygosity are confirmed by orthogonal methods (Sanger, MLPA, qPCR).
CNV SENSITIVITY	NGS-based copy number variations (CNV) are detected with a sensitivity of above 95% for all homozygous deletions and heterozygous deletions/duplications spanning at least three consecutive exons. Heterozygous CNVs spanning less than three exons cannot reliably be detected, are therefore excluded from routine analysis, and will only be inspected and reported upon medical or technical indication.
REPORTING	Pathogenic and likely pathogenic variants are reported following American College of Medical Genetics and Genomics (ACMG) classification guidelines. Variants of uncertain significance (VUS) are not reported in any of the following cases: the described phenotype(s) is explained by detected pathogenic or likely pathogenic variant(s); the detected VUS are not related to the described phenotype(s) of the patient or family members; and in the lack of sufficient clinical information.
TAT	25 business days*

SNVs: single nucleotide variants; InDels: small insertions/deletions; CNVs: copy number variations; MLPA: Multiplex ligation-dependent probe amplification; qPCR: quantitative polymerase chain reaction.

* For relevant panels where a quick medical answer is needed we have shorter TAT (10 or 15 days) please visit www.centogene.com/diagnostics/ngs-panels for more information

Going The Extra Mile

All of our NGS panels include sequencing, deletion/duplication (CNV) analysis, and complementary assays to offer the most complete analysis for maximum diagnostic yield.

DELETION/DUPLICATION	High resolution NGS-based CNV analysis to detect larger deletions and duplications is included in all our panels at no extra cost. Deletion/duplications constitute 5 – 10% of disease-causing variants. By including CNV analysis in our panels, the potential of providing the most accurate diagnosis increases.
COMPLEMENTARY ASSAYS	To maximize clinical utility, our panels are reinforced with auxiliary assays such as repeat expansions, MLPA, or Sanger Sequencing to cover genes/regions that cannot be examined by current sequencing technology.
IMPROVED INTERPRETATION	Our rare disease-centric Bio/Databank enables access to more than 30 million unique variants for best medical interpretation.
VARIANT RECLASSIFICATION PROGRAM	All our panels are automatically entered into our variant reclassification program. This program supports the identification of new genetic evidence, and physicians will be notified free of charge for life if the nature of a previous diagnosis has been impacted.