

NGS Panels Pediatric Neurology

A Targeted Approach For Testing
Neurological Disorders

PRODUCT SHEET

NGS Panels For Pediatric Neurology

CENTOGENE's selection of Next Generation Sequencing (NGS) gene panels for hereditary neurological disorders (HNDs) can help pediatricians pinpoint the genetic cause of major neurological disorders in children including movement disorders, epilepsy, autism spectrum disorders, neuropathies, neuromuscular disorders and others.

Our panels follow a phenotype-directed approach that includes all relevant clinical genes and genes necessary for the 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, minimizes the risk of incorrect 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



Powered by CENTOGENE's Biodatabank, the **world's largest real-world data repository** for rare and neurodegenerative diseases



High-quality analysis for precise clinical interpretation using advanced bioinformatics and artificial intelligence-powered tools

NGS Panel Options

PEDIATRIC NEUROLOGY PANELS	ASSOCIATED DISEASE(S)
CentoMito® Comprehensive	Panel includes syndromes related to mutations in mitochondrial genes and nuclear genes related to mitochondrial diseases such as chronic progressive external ophthalmoplegia, Kearns-Sayre syndrome, lactic acidosis, Leigh’s syndrome, mitochondrial disorders, mitochondrial encephalomyopathy with ragged red fibers, myogastrointestinal encephalomyopathy, NARP, neonatal mitochondrial hepatopathies, Pearson syndrome and stroke-like episodes.
CentoMito® Genome	Panel includes syndromes related to mutations in mitochondrial DNA such as chronic progressive external ophthalmoplegia, Kearns-Sayre syndrome, leber hereditary optic neuropathy, Leigh’s-like syndrome, Leigh’s syndrome, mitochondrial disorders, and NARP.
CentoIEM	Inborn Errors of Metabolism largely impact human diseases. CentoIEM is a metabolic and liver disease gene panel that covers a large array of different disorders and includes genes responsible for diverse phenotypes such as Aicardi-Goutieres syndrome, amino-acidopathies, ceroid lipofuscinosis, congenital glycosylation disease, familial hypercholesterolemia, fatty liver disease, Leigh’s syndrome, liver cirrhosis, mitochondrial encephalopathy, neurodegeneration with brain iron accumulation, organic acidurias, Refsum disease, porphyrias and urea cycle disorders, among others. Panel includes mtDNA analysis.
CentoMetabolic MOx	Our panel integrates genetic and biochemical testing to test for patients suspected of having a metabolic disorder of presenting complex, overlapping symptoms, a metabolic crisis, or neurological conditions of unknown etiology. Panel includes complementary testing by proprietary biomarkers and enzyme-activity assays if applicable.
CentoNeuro	Our largest panel is designed to detect a wide array of neurological disorders including but not limited to arthrogryposis multiplex congenita, dystonia, epilepsy, familial hemiplegic migraine, hypogonadotropic hypogonadism, Kallman syndrome, Leigh’s syndrome, leukodystrophy, Meckel syndrome, mitochondrial encephalomyopathy, neonatal mitochondrial hepatopathies, and neuromuscular disorders between others.
Epilepsy	Our epilepsy panel covers different types of seizure syndromes including but not limited to Aicardi-Goutieres syndrome, brain iron accumulation syndromes, Dravet syndrome, early infantile epileptic encephalopathy, epilepsy partial, epilepsy generalized, epilepsy absence, myoclonic epilepsy, epileptic encephalopathy, Leigh’s syndrome, leukodystrophy, mitochondrial encephalo-myopathy, mitochondrial DNA depletion, peroxisome biogenesis disorders, hypomagnesemia and urea cycle disorders. Panel includes repeat expansion analysis for <i>CSTB</i> and mtDNA analysis.
Intellectual Disability	Panel covers neurodevelopmental disorders such as Aicardi-Goutieres syndrome, epileptic encephalopathy, intellectual disability, microcephaly, neural migration disorder, neurodevelopmental disorders and syndromic autism. Panel includes repeat expansion analysis for <i>FMR1</i> and mtDNA analysis.
Neuromuscular	Our panel for muscular diseases covers disorders such as Charcot-Marie-Tooth, congenital myasthenic syndromes, congenital myopathies, metabolic myopathies, muscular dystrophies, myofibrillar myopathies, nemaline myopathies, spinal muscular atrophy type 1 and other syndromes with hypotonia, muscular weakness or myotonia. Panel includes repeat expansion analysis for <i>DMPK</i> , E7 homozygous deletion screening for <i>SMN1</i> , and mtDNA analysis.

Key Features and Performance

COVERAGE

- $\geq 99.0\%$ targeted regions covered at $\geq 20x$
- For each gene, all SNVs described in HGMD® and CENTOGENE's Biodatabank are covered, including relevant deep intronic and regulatory variants.

GENES

For an overview of included genes, please visit our NGS Panel webpage:
centogene.com/diagnostics/ngs-panels

SPECIFICITY

$\geq 99.9\%$ 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; in the lack of sufficient clinical information.

TAT

25 business days

Going the Extra Mile

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 to cover genes/regions that cannot be examined by current sequencing technology.

MITOCHONDRIAL GENOME

High-quality mitochondrial testing is now included for panels where symptoms may be caused by mitochondrial DNA mutation.

IMPROVED INTERPRETATION

CENTOGENE's Biodatabank, the world's largest real-world data repository for rare and neurodegenerative diseases enables access to more than 31 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.