

Our NGS Panels

A TARGETED APPROACH FOR
TESTING GENETIC DISORDERS

Next Generation Sequencing (NGS) Panels



Diagnosing rare disease patients can be difficult, even for the most experienced physicians. With our phenotype driven NGS panels, we make diagnostics easy by screening for multiple diseases in a single test. Thanks to our selection of clinically relevant genes, carefully chosen to maximize the diagnostic yield and allow a differential diagnosis, as well as our inclusive copy number variant (CNV) analysis, our patients receive the most extensive screenings.

Our specialized NGS Panel portfolio tests for a wide selection of hereditary genetic disorders across 16 different diseases categories, including neurology, endocrinology, cardiovascular, dysmorphology, and metabolic disorders. Our unique approach, which screens for syndromes with overlapping phenotype(s), maximizes clinical utility, de-risks panel choice, and increases cost-effectiveness – ultimately simplifying and shortening the diagnostic odyssey for rare disease patients.

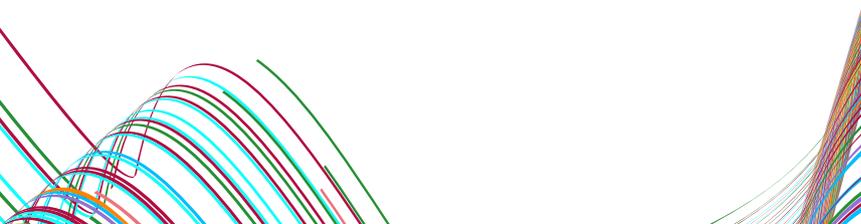
When choosing one of our NGS panels, you and your patients can rest assured that you are receiving the highest quality sequencing supported by best-in-class data analysis and comprehensive medical reports powered by insights from over half a million patients.

When to choose a panel?

Recommended when patients meet any of the following criteria*:

- Genetically heterogeneous disorders where a similar manifestation might occur through different genetic mechanisms
- Phenotypes corresponding to disturbances of the same pathway
- Disorders sharing one manifestation with varying presentations
- Disorders with overlapping manifestations corresponding to differential diagnosis
- Disorders where multiple genes are linked to the condition
- Family history suggestive of a genetic condition, but without a precise diagnosis

* Genet Med. 2015 Jun;17(6):444-51. doi: 10.1038/gim.2014.122. Epub 2





Panel Features

Coverage: ≥99.5% targeted regions covered at ≥20x

CNV analysis: Included in our panels at no extra cost

Variants: All single nucleotide variants described in HGMD and CentoMD®, including relevant deep intronic and regulatory variants

Clinical Information: Detailed and specific clinical information (i.e., HPOs) is required for variant interpretation and medical diagnosis

Reporting: Pathogenic and likely pathogenic variants are reported following ACMG classification guidelines and according to the clinical information provided. Variants of uncertain significance (VUS) related to the described phenotype(s) of the patient or family members are reported only if the described phenotype(s) is not explained by detected pathogenic or likely pathogenic variant(s). VUS are not reported when there is insufficient clinical information or in our Oncogenetic panels.

Tabular List: In addition to primary findings directly associated with the patient's phenotype, CENTOGENE provides extra information regarding potentially relevant pathogenic and likely pathogenic variants found in your patient's sample*. Our list makes often unreachable information accessible to physicians/genetic counsellors – allowing them to take actionable steps to potentially help prevent morbidity and mortality of the patient and/or their family.

Complementary Assays: Our panels are reinforced when necessary assays to overcome potential limitations of sequencing.

TAT: 25 days

Some exceptions may apply. For complete information about our panels, please visit:
<https://www.centogene.com/diagnostics/ngs-panels.html>

* This list differs from the ACMG list of incidental findings (Genetics in Medicine volume 21, pages 1467–1468(2019)) for more details please visit <https://www.centogene.com/diagnostics/medical-reporting/p-lp-gene-reporting.html>

The CENTOGENE Advantage

A comprehensive diagnostic solution beyond DNA testing

OUR DIAGNOSTIC SERVICES ARE MORE THAN LABORATORY AND BIOINFORMATICS.

CentoCard®

Our quick, cost-effective, and hassle-free solution for shipment of clinical blood samples for genetic testing. Collected samples are unaffected by shipping time and temperature, and a single card enables genetic and metabolic testing.

Extended Phenotyping

Structuring your patient's symptoms into Human Phenotype Ontology (HPO) terms ensures the best quality of clinical information for data interpretation.

Data Safety and Research Use

With transparent and easy-to-understand consent forms, your patients can make educated decisions without worrying about data protection. By consenting to the research and storage option, you and your patients will advance research, the understanding of rare diseases, and the quality of future diagnoses and therapies.

Multomics Testing

Continuous research identifies and validates biomarkers, increasing disease understanding and enabling therapy monitoring. This has already added diagnostic certainty to lysosomal storage disorders and other diseases.

CentoPortal®

A user-friendly and fully secure online service designed to assist in ordering tests, transferring patient data, administering patients' samples, and accessing your diagnostic reports 24/7. Please go to: www.centoportal.com

CentoMD®

Our extensive rare disease data repository with over 400,000 analyzed case and more than 12 million unique variants supports our world-class medical interpretation.

Variant Reclassification Program

CENTOGENE has a highly robust and ongoing variant reclassification program based on new genetic evidence. If re-classification affects the nature of the genetic diagnosis of the patient, physicians will be notified free of charge.

World-Class Expertise

CENTOGENE's reputation is built on an international team of genetic and bioinformatics experts, the latest lab technology, continuously improved processes and protocols, and unique data analysis software.



OUR COMMITMENT:

We have a life-long commitment to our patients to make rare diseases 'un-rare' – using our global impact, expertise, and passion to provide world-class genetic knowledge and accelerate orphan drug development.

Your partner of choice

For further information and support, please contact our closest representative or our customer support team, easily accessible by phone or email.

www.centogene.com

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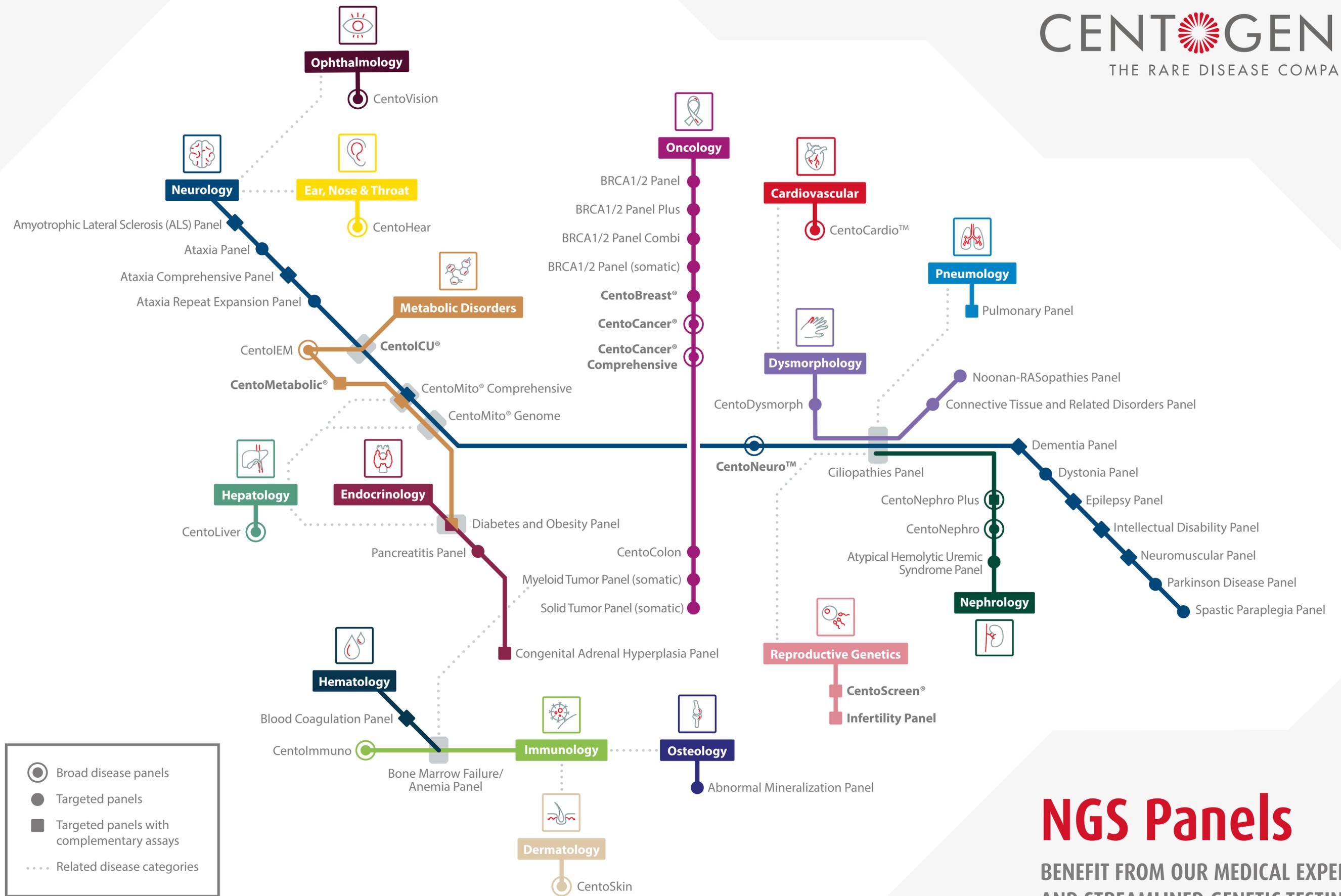
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NGS Panels

BENEFIT FROM OUR MEDICAL EXPERTISE
AND STREAMLINED GENETIC TESTING