CENTOGENE
THE RARE DISEASE COMPANY

NGS Panels

CentoMetabolic MOx

More Answers Today. More Options Tomorrow.

CentoMetabolic MOx

Inherited Metabolic Disorders (IMDs) are a group of rare conditions caused by genetic defects that disrupt the cellular metabolism. A growing number of IMDs are treatable if diagnosed early, but can be quickly fatal without prompt identification. With a multiomic approach, we can help you and your patients to accelerate the critical journey from symptoms to diagnosis by avoiding stepwise testing – saving time, resources, and pivotal years amid often rapid IMD progression.

CENTOGENE's multiomic panel – CentoMetabolic MOx – has been designed to test for a wide range of IMDs – integrating genetic and biochemical testing, including enzyme assays as well as a selection of proprietary biomarkers. When genetic variants relevant to your patient are detected via CentoMetabolic MOx, we will automatically complement the analysis with biomarker and/or enzyme testing (if applicable) and include the results in your medical report. In addition, CentoMetabolic MOx includes an evaluation of Copy Number Variants (CNV) at no extra cost. CentoMetabolic MOx gives you the confidence of a complete clinical picture, while laying the roadmap to personalized treatment options.

The CENTOGENE Advantage

Multiomic panel integrating genetic and biochemical testing in a single **solution, for fast and accurate diagnosis** of a wide range of rare inherited metabolic disorders

Best in class insights powered by CENTOGENE's Biodatabank, the world's largest real-world data repository for rare and neurodegenerative diseases

Biochemical testing to support variant classification, leading to **higher diagnostic yields**

Dedicated team of medical experts to provide best **clinical interpretation and life-long support**

Who Should Consider CentoMetabolic MOx?

Physicians providing treatment for patients matching any of the following criteria:

- Suspected metabolic disorder
- Infants with lethargy or abdominal pain or vomitting or jaundice or metabolic acidosis
- Developmental delay
- Abnormal newborn screening results
- Admission to a neonatal intensive care unit (NICU), especially due to epilepsy of unclear origin and disturbed consciousness
- Symptoms related to neurological conditions of unknown etiology

What Genes and Disorders Are Targeted?

CentoMetabolic MOx targets close to 180 IMDs. The content and design of the panel is based on our continuously enhanced medical expertise and knowledge of rare metabolic disorders, including the latest medical and in-house findings.

The table below shows the distribution of genes and targeted metabolic disorders depending on 18 different disease categories:

Explore All Covered Genes Online

Type of Metabolic Disorders Covered	# Genes*
Congenital disorders of glycosylation and other disorders of protein modification	2
Defects in cholesterol and lipoprotein metabolism	2
Defects in hormone biogenesis or function	7
Disorder of phosphate, calcium, and vitamin D metabolism	3
Disorders in the metabolism of purines, pyrimidines, and nucleotides	6
Disorders in the metabolism of trace elements and metals	6
Disorders in the metabolism of vitamins and (non-protein) cofactors	10
Disorders of amino acid and peptide metabolism	33

Type of Metabolic Disorders Covered	# Genes*
Disorders of carbohydrate metabolism	35
Disorders of energy metabolism	6
Disorders of fatty acid and ketone body metabolism	3
Disorders of lipid and lipoprotein metabolism	8
Disorders of neurotransmitter metabolism	1
Disorders of porphyrin and heme metabolism	8
Disorders of the metabolism of sterols	16
Lysosomal disorders	48
Peroxisomal disorders	16
Porphyria and bilirubinemia	1

* Due to overlapping phenotypes, particular genes are listed in more than one category as they are associated with more than one disorder.

Key Features and Performance

Multiomic Approach	198 genes, and over 20 enzymes and biomarkers associated with more than 180 IMDs
Coverage	• ≥ 99.5 % targeted regions covered at ≥ 20 x • For each gene, all clinically relevant variants described in HGMD® and CENTOGENE's Biodatabank are covered, including deep intronic and regulatory variants
Specificity	≥ 99.9 % guaranteed for all reported variants. Variants with low quality and / or unclear zygosity are confirmed by orthogonal methods (Sanger, MLPA, qPCR)**
Material	≥ 1 CentoCard® or 4 ml EDTA blood
TAT	15 business days

** **MLPA:** Multiplex ligation-dependent probe amplification; **qPCR:** quantitative polymerase chain reaction.

Beyond Genetics with CENTOGENE's Biomarker and Enzyme Testing

CentoMetabolic MOx includes biomarkers and enzymatic assays for over 20 metabolic disorders. Biomarkers serve as measurable indicators of pathological processes. They are typically directly linked to genetic variants in specific genes and can predict, diagnose, monitor, and assess the severity of a disease. Measuring the cellular activity of an enzyme can also be used as a tool for the diagnosis and monitoring of a disease, as well as treatment efficacy.

Our multiomic- and big data-based approaches allow us to continuously discover new highly specific biomarkers. All new biomarkers and biochemical assays clinically relevant for metabolic disorders will be included in this panel, advancing the understanding of metabolic disorders, accelerating the path from diagnosis to personalized treatment.

Biomarkers and Enzymes Included in Multiomic Portfolio

In the multiomic testing environment, biochemical testing evaluates the impact of potentially clinically relevant variants (Variants of Uncertain Significance, Likely Pathogenic, and Pathogenic Variants) in genes for the following analytes.

Biomarkers

Gaucher disease
Glucosylsphingosine (lyso-Gb1)*

Fabry disease
Lyso-ceramide trihexoside (lyso-Gb3)

Niemann-Pick disease type A / B / C
Lyso-SM-509

Aromatic L-amino acid decarboxylase (AADC) deficiency
3-O-methyldopa (3-OMD)

Enzymes

Neuronal Ceroid Lipofuscinosis

Santavuori-Haltia disease
Palmitoyl-protein- thioesterase

Jansky-Bielschowsky disease
Tripeptidyl-peptidase

Enzymes

Oligosaccharidoses and Sphingolipidoses

Wolman disease
Acid lipase

Pompe disease
Alpha-glucosidase

Fucosidosis
Alpha-fucosidase

Fabry disease
Alpha-galactosidase

Schindler / Kanzaki disease
Alpha-N-acetylgalactosaminidase

Gaucher disease
Beta-glucocerebrosidase

Tay-Sachs disease
Beta-hexosaminidase

Beta-mannosidosis
Beta-mannosidase

Sandhoff disease
Total-hexosaminidase

Enzymes

Mucopolysaccharidosis

Hurler syndrome (MPS I)
Alpha-L-iduronidase

Hunter syndrome (MPS II)
Iduronate-2-sulfatase

Sanfilippo syndrome B (MPS III B)
Alpha-N-acetylglucosaminidase

GLB1-Related Disorders
Beta-galactosidase

Maroteaux-Lamy syndrome (MPS VI)
Arylsulfatase B

Sly syndrome (MPS VII)
Beta-glucuronidase

* A method using Lyso-Gb1 is covered by US Patent No.10,859,580, other pending US applications, and pending applications and patents in other jurisdictions

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