CENTOGENE
THE RARE DISEASE COMPANY

NGS Panels

A Targeted Approach for Testing Genetic Disorders

Working together...

Benefit from our medical expertise and streamlined genetic testing

CENTOGENE is fully committed to bring the best possible diagnostic solutions to patients and their families. We strive to incorporate the latest in-house findings and medical research in our products to improve and ease the diagnostic odyssey of rare disease patients. Our Next Generation Sequencing (NGS) panels are designed to reflect the fast-growing knowledge of complex associations of genes with diseases as well as maximize clinical sensitivity.

The gene composition of each panel has been revised to include the latest gene discoveries and to provide the highest clinical certainty. Our comprehensive phenotype-directed diagnostic panels include all relevant genes necessary for differential diagnosis of syndromes with overlapping phenotypes – enabling the diagnosis of diseases that could have been missed otherwise. This enhancement increases clinical utility, de-risks panel choice, increases cost-effectiveness, and ultimately simplifies the diagnostic process.

When choosing one of our NGS panels, your patients will receive high-quality sequencing, best-in-class data analysis and interpretation, as well as comprehensive medical reports. As always, our Customer Support team is available to answer questions and help you during the diagnostic process in any way we can.

Explore Our NGS Panel Portfolio

Disclaimer: Due to continous developments in our product portfolio the gene numbers in our panels are subject to change without prior notice. Lists of common syndromes and disorders covered is not exhaustive. For the most up-to-date list of included genes and corresponding phenotypes, please visit: centogene.com/diagnostics/ngs-panels

Panel Specifications

Coverage	The target region for each gene comprises all exons; ±10 bp flanking regions; known pathogenic and likely pathogenic variants described in HGMD® and CENTOGENE's Biodatabank, including relevant deep intronic and regulatory variants, which were known at the time of the assay design.
Specificity	≥99.9% guaranteed for all reported variants. Variants with low quality and/or unclear zygosity are confirmed by orthogonal methods (Sanger, MLPA, qPCR).
Complementary Assays	Some 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.
Genes	For a complete overview of included genes, please visit: centogene.com/diagnostics/ngs-panels
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.
Mitochondrial Genome	High-quality mitochondrial testing is now included for panels where symptoms may be caused by mitochondrial DNA mutation.
Medical Reporting	Pathogenic and likely pathogenic variants are reported following ACMG classification guidelines.Variants of uncertain significance (VUS) are not reported in any of the following cases: The described phenotype is explained by detected pathogenic or likely pathogenic variants; the detected VUS are not related to the described phenotype of the patient or family members; lack of sufficient clinical information; in oncogenetic panels.
Requested Material	For more details of accepted materials please visit: centogene.com/diagnostics/how-to-order

Disease Categories

CentoCardio

Genes: 327

CentoCardio includes the most relevant genes for arrythmias, congenital heart disease, and cardiomyopathies. Syndromes included: Long and short QT syndrome, Brugada syndrome, catecholaminergic polymorphic ventricular tachycardia, cardiomyopathies dilated and hypertrophic, and congenital heart defects. In addition, this panel includes vascular abnormalities, such as dolichoectasia and hereditary hemorragic telangiectasia.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Arrhythmogenic right ventricular cardiomyopathy
- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Congenital heart defects
- Dilated cardiomyopathy
- Dolichoectasia
- Hereditary arrhythmia syndromes
- Hereditary hemorrhagic telangiectasia
- Heterotaxy syndrome
- Hypertrophic cardiomyopathy
- Hypomagnesemia
- Long QT syndrome
- Short QT syndrome

CentoSkin

Genes: 160

CentoSkin is our diagnostic test for patients displaying skin disorders. Our panel includes genes for hypotricosis, epidermolysis bullosa, and congenital ichthyosis, among others. In addition, CentoSkin tests for albinism and other conditions with similar pigmentation abnormalities such as Hermasky-Pudlak syndrome, Griscelli snydrome and Waardenburg syndrome. For melanoma, please check our oncology panels.

TAT	25 business days
Coverage	≥99.0% ≥20x
Details	CNV analysis included

Common Syndromes & Disorders Covered

- Albinism oculocutaneous
- Chediak-Higashi syndrome
- Congenital ichthyosis
- Cutis laxa
- Epidermolysis bullosa
- Griscelli syndrome
- Hermasky-Pudlak syndrome
- Ichthyosis extended
- Non-syndromic hypotrichosis
- Waardenburg syndrome

CentoDysmorph

Genes: 776

CentoDysmorph is designed to help physicians diagnose patients that suffer from a dysmorphic syndrome. The panel includes craniosynostosis, craniofacial disorders, cleft / lip palate, holoprosencephaly, Waardenburg syndrome, Hirschsprung disease, lissencephaly, and brain malformation disorders, among others.

Additionally, CentoDysmorph includes genes related to RASopathies. RASopathies are a group of genetic syndromes caused by germline mutations in genes that encode components or regulators of the RAS / mitogen-activated protein kinase (MAPK) pathway. This panel includes genes related to neurofibromatosis type 1, Noonan syndrome, Noonan syndrome with multiple lentigines, capillary malformation-arteriovenous malformation syndrome, cardiofaciocutaneous syndrome, Costello syndrome, and Legius syndrome, among others.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Bardet-Biedl syndrome
- Cardiofaciocutaneous syndrome
- Cerebral cavernous malformations
- Ciliopathies
- Cleft lip and palate
- Coffin-Siris syndrome
- Cornelia de Lange syndrome
- Ciliopathic skeletal dysplasias
- Craniosynostosis and craniofacial disorders
- Heterotaxy syndrome
- Hirschsprung disease
- Holoprosencephaly
- Klippel-Feil syndrome
- Lissencephaly and brain malformation
- Meckel syndrome
- Metaphyseal dysplasia
- Micro syndrome
- Microphthalmia / anophthalmia / coloboma spectrum
- Multiple epiphyseal dysplasia
- Neurofibromatosis
- Noonan-RASopathies syndromes
- Seckel syndrome
- Skeletal dysplasia extended
- Stickler syndrome
- Tuberous sclerosis
- Waardenburg syndrome

Connective Tissue and Related Disorders Panel

Genes: 76

Our Connective Tissue and Related Disorders Panel provides a one-step evaluation of several genes to detect different disorders with similar phenotypes, such as Marfan syndrome, Loeys-Dietz syndrome, cutis laxa, Ehlers-Danlos snydrome, Stickler syndrome, and familial thoracic aortic aneurysm and dissection.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Cutis laxa
- Ehlers-Danlos syndrome
- Familial thoracic aortic aneurysm and dissection
- Loeys-Dietz syndrome
- Marfan syndrome
- Osteogenesis imperfecta
- Stickler syndrome

CentoHear

Genes: 233

Hearing loss is a common condition in children, affecting 1 in 100 live births. In more than 50 % of cases, there is a genetic cause for this disorder, of which 70 % experience non-syndromic hearing loss.

CentoHear includes genes associated with syndromic and non-syndromic hearing loss. Both autosomal recessive and dominant genes are included in the panel. In addition, CentoHear includes several other syndromes, such as Alport syndrome, Pendred syndrome, Waardenburg syndrome, Usher syndrome, and branchiootorenal, among others.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Alport syndrome
- Coffin-Lowry snydrome
- Deafness autosomal recessive and dominant
- Non-syndromic hearing loss
- Pendred syndrome
- Perrault syndrome
- Pfeiffer syndrome
- Sensorineural hearing loss
- Stickler syndrome
- Syndromic hearing loss
- Usher syndrome
- Waardenburg syndrome
- Wolfram syndrome

Congenital Adrenal Hyperplasia (CAH) Panel

Genes: 12

Our Congenital Adrenal Hyperplasia (CAH) Panel is designed for patients suspected of having CAH. CAH is a group of inherited disorders characterized by improper functioning of the adrenal glands, leading to abnormal production of steroid hormones, as cortisol or aldosterone. Our panel includes analysis of the *CYP21A2* gene, which codes for the enzyme 21-hydroxylase. More than 90 % of CAH cases are caused by a deficiency of this enzyme.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included MLPA: *CYP21A2* Sanger sequencing: *CYP21A2*

Common Syndromes & Disorders Covered

- Congenital adrenal hyperplasia
- Pigmented nodular adrenocortical disease

Diabetes and Obesity Panel

Genes: 265

Our Diabetes and Obesity Panel is recommended for patients with abnormalities in glucose metabolism, such as hyperinsulinemic hypoglicemia, diabetes neonatal, Maturity-onset diabetes of the young (MODY), diabetes in adults, and familial hypercholesterolemia as well as for patients displaying insulin resistance, from mild to the severe spectrum (Donohue syndrome), and for patients with familial hyperinsulinism. Prader-Willi syndrome can be detected with this panel, however, other disorders caused by imprinting errors or uniparental disomy such as 6q24-related transient neonatal diabetes mellitus and Beckwith-Wiedemann syndrome are not included in this panel.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included MLPA: 15q11

Common Syndromes & Disorders Covered

- Bardet-Biedl syndrome
- Congenital glycosylation disease
- Congenital hyperinsulinism
- Congenital hypothyroidism
- Diabetes insipidus
- Growth hormone deficiency
- Familial hypercholesterolemia
- Hypoglycemia
- Maturity-onset diabetes of the young
- Neonatal diabetes
- Obesity
- Prader-Willi syndrome

→ also view: Metabolic Disorders p.22

Pancreatitis Panel

Genes: 29

Our Pancreatitis Panel includes genes associated with chronic pancreatitis and additional genes for differential diagnosis; genes associated with pancreatic cancer are also included.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included Sanger sequencing: *CFTR*, exon 10

Common Syndromes & Disorders Covered

- Pancreatic cancer
- Pancreatitis

Blood Coagulation Panel

Genes: 112

Our Blood Coagulation Panel contains genes to diagnose thrombophilia, thrombocytopenia, hereditary hemorrhagic telangiectasia, Arthrogryposis-renal dysfunctioncholestasis (ARC) syndrome , Hermasky-Pudlak syndrome, coagulation factor disorders, hemophilia, and platelet related disorders.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included Targeted mutation analysis for *F8* to detect inversion of intron 1 and intron 22A included

Common Syndromes & Disorders Covered

- Afibrinogenemia
- Arthrogryposis-renal dysfunction-cholestasis syndrome
- Coagulation factor disorders
- Hemophilia
- Hereditary angioedema
- Hereditary hemorrhagic telangiectasia
- Hermasky-Pudlak syndrome
- Platelet related disorders
- Shwachman-Diamond syndrome
- Thrombocytopenia
- Thrombophilia

Bone Marrow Failure / Anemia Panel

Genes: 214

Our Bone Marrow Failure / Anemia Panel is intended for patients with abnormalities in more than two blood cell types (red blood cell, white blood cell, and platelets) who present symptoms of lethargy, recurrent infections, excessive bleeding, abnormal pigmentation, enlarged spleen, and malignancies. Some specific disorders detected with this panel are hemophagocytic lymphohistiocytosis, Seckel syndrome, thrombocytopenia, Fanconi anemia, dyskeratosis congenita, Shwachman-Diamond syndrome as well as other types of anemias, such as thalassemia alpha and beta, sickle cell disease, spherocytosis, megaloblastic anemia, congenital sideroblastic anemia, and dyserythropoietic anemia.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Bleeding disorders
- Bone marrow failure syndrome
- Congenital dyserythropoietic anemia
- Congenital sideroblastic anemia
- Diamond-Blackfan anemia
- Fanconi anemia
- Hemolytic anemias
- Hemophagocytic lymphohistiocytosis
- Hereditary spherocytosis
- Megaloblastic anemia
- Seckel syndrome
- Shwachman-Diamond syndrome
- Sitosterolemia
- Thrombocytopenia

→ also view: Immunology p.16

Bone Marrow Failure / Anemia Panel

Genes: 214

Our Bone Marrow Failure / Anemia Panel is intended for patients with abnormalities in more than two blood cell types (red blood cell, white blood cell, and platelets) who present symptoms of lethargy, recurrent infections, excessive bleeding, abnormal pigmentation, enlarged spleen, and malignancies. Some specific disorders detected with this panel are hemophagocytic lymphohistiocytosis, Seckel syndrome, thrombocytopenia, Fanconi anemia, dyskeratosis congenita, Shwachman-Diamond syndrome as well as other types of anemias, such as thalassemia alpha and beta, sickle cell disease, spherocytosis, megaloblastic anemia, congenital sideroblastic anemia, and dyserythropoietic anemia.

TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Bleeding disorders
- Bone marrow failure syndrome
- Congenital dyserythropoietic anemia
- Congenital sideroblastic anemia
- Diamond-Blackfan anemia
- Fanconi anemia
- Hemolytic anemias
- Hemophagocytic lymphohistiocytosis
- Hereditary spherocytosis
- Megaloblastic anemia
- Seckel syndrome
- Shwachman-Diamond syndrome
- Sitosterolemia
- Thrombocytopenia

→ also view: Hematology p.15

CentoImmuno

Genes: 441

Our CentoImmuno panel is specifically designed to target genes associated with Human Inborn Errors of Immunity (IEI). The later consists of a large and diverse group of disorders presenting a common clinical characteristic, suppressed innate and adaptive immunity. Causative variants of genes included in CentoImmuno most often confer susceptibility to infectious disease, autoinflammatory disease, neoplasia, autoimmunity, or allergies.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Anhidrotic ectodermal dysplasia with immunodeficiency
- Autoinflammatory disorders
  Recurrent fever, type 1 interferonopathies, sterile inflammation
- Bone marrow failure
  Fanconi anemia, dyskeratosis congenita, bone marrow failure syndrome
- Common variable immunodeficiency
- Complement deficiencies
  atypical hemolytic uremic syndrome
- Congenital defects of phagocyte
  Neutropenia, Shwachman-Diamond Syndrome, functional defects
- Congenital thrombocytopenia
- Defects of intrinsic and innate immunity
  predisposition to viral, bacterial, fungal and parasitic infections
- Defects of vitamin B12, folate metabolism
- Diseases of immune dysregulation
  Hemophagocytic lymphohistiocytosis, EBV susceptibility
- DNA repair defects
- Hyper IgE syndromes
- Immunodeficiencies affecting cellular and humoral immunity
  (Severe) combined immunodeficiencies
- Immunoosseous dysplasias
- Mendelian susceptibility to mycobacterial disease
- Predominantly antibody deficiencies
  hypogammaglobulinemia, other antibody deficiencies
- Syndromes with autoimmunity
- Thymic defects with additional congenital anomalies

CentoICU®

Genes: 856

CentoICU® is a comprehensive NGS Panel that includes genes explicitly selected for the genetic testing of critically ill newborns and children under 24 months in intensive care units (ICU). It is designed to address multiple genetic conditions that may be present in the newborn or early childhood period, with many having overlapping phenotypes and immediate implications for treatment initiation. It allows clinicians to utilize a single test to provide an accurate diagnosis of newborn-related diseases using dried blood spots.

TAT	15 business days FAST: 10 business days
Coverage	≥99.0% ≥20x

Common Syndromes & Disorders Covered

- Alagille syndrome
- Alpha-Thalassemia
- Arginase deficiency
- Beta-Thalassemia
- Biotinidase deficiency
- Biotin-thiamine-responsive basal ganglia disease
- Carnitine deficiency
- Congenital hypothyroidism
- Cystic Fibrosis
- Dystonia DOPA responsive
- Factor VII deficiency
- Glucose transporter 1 deficiency
- Glutaric acidemia type 1
- Hereditary fructose intolerance
- Holocarboxylase synthetase deficiency
- Maple syrup urine disease (MSUD)
- Nonketotic hyperglicinemia
- Phenylketonuria
- Pompe disease
- Primary coenzyme Q10 deficiency
- Pyridoxamine 5 phosphate oxidase deficiency
- Pyridoxine-dependent epilepsy
- Pyruvate carboxylase deficiency
- Tuberous sclerosis complex
- Tyrosinemia type I
- VLCAD deficiency

→ also view: Neurology p.26

CentoIEM

Genes: 744

Inborn Errors of Metabolism (IEM) largely impact human diseases. CentoIEM is a metabolic and liver disease gene panel that screens for an array of different disorders and contains genes responsible for diverse phenotypes, including intermediary metabolism, such as aminoacidopathies, organic acidurias, urea cycle disorders, sugar intolerance, mental disorders, and porphyrias, among others. Genes linked to cytoplasmic and mitochondrial energetic processes and metabolism affecting cellular organelles, such as lysosomal, peroxisomal, glycosylation, and cholesterol synthesis are also included.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Aicardi-Goutieres syndrome
- Ceroid lipofuscinosis
- Congenital glycosylation disease
- Familial hypercholesterolemia
- Fatty acid oxidation disorder
- Fatty liver disease
- Glycogen storage disease
- Hemophagocytic lymphohistiocytosis
- Hereditary hemochromatosis
- Hereditary spherocytosis
- Leigh syndrome and mitochondrial encephalopathy
- Leukodystrophy and peroxisome biogenesis disorders
- Lipodystrophy syndromes
- Liver cirrhosis
- Lysosomal storage disease
- Mucopolysaccharidosis
- Neurodegeneration with brain iron accumulation
- Nonketotic hyperglycinemia
- Organic acidemias
- Porphyria
- Refsum disease
- Urea cycle disorder

CentoMetabolic® MOx

Genes: 206

CentoMetabolic® MOx was developed specifically for patients suspected of having a metabolic disorder or presenting complex, overlapping symptoms, a metabolic crisis, or neurological conditions of unknown etiology. It provides short turnaround times – targeting critically ill patients in NICU/PICU. It leverages a multiomic approach by including enzyme activity testing where applicable, as well as a proprietary selection of biomarkers that is continuously updated.

TAT	15 business days
Coverage	≥99.5% ≥20x
Methods	CNV analysis included Complementary biochemical testing by proprietary biomarkers and enzyme-activity assays if applicable

Common Syndromes & Disorders Covered

- Congenital disorders of glycosylation and other disorders of protein modification
- Defects in cholesterol and lipoprotein metabolism
- Defects in hormone biogenesis or function
- Disorder of phosphate, calcium and vitamin D metabolism
- Disorders in the metabolism of purines, pyrimidines and nucleotides
- Disorders in the metabolism of trace elements and metals
- Disorders in the metabolism of vitamins and (non-protein) cofactors
- Disorders of amino acid and peptide metabolism
- Disorders of carbohydrate metabolism
- Disorders of energy metabolism
- Disorders of fatty acid and ketone body metabolism
- Disorders of lipid and lipoprotein metabolism
- Disorders of neurotransmitter metabolism
- Disorders of porphyrin and heme metabolism
- Disorders of the metabolism of sterols
- Lysosomal disorders
- Peroxisomal disorders
- Porphyria and bilirubinemia

CentoMito Comprehensive

Genes: 451

CentoMito Comprehensive covers the entire mitochondrial genome with detection of heteroplasmy down to 15% and tests for nuclear genes related to mitochondrial diseases. Mitochondrial diseases are genetic conditions that occur when mitochondria fail to produce enough energy for the cell. Genetic mutations related to mitochondria cause symptoms mainly in organs, that consume large amounts of energy. These organs include the eye, kidney, pancreas, blood, inner ear, colon, skeletal muscle, heart, and brain.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included ≥15.0% mitochondrial heteroplasmy can be confidently detected

Common Syndromes & Disorders Covered

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leigh syndrome and maternally inherited Leigh syndrome
- Mitochondrial disorders
- Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes
- Myoclonus epilepsy with ragged red fibers
- Myogastrointestinal encephalomyopathy
- Neurogenic muscle weakness, ataxia and retinitis pigmentosa (NARP)
- Neonatal mitochondrial hepatopathies
- Pearson syndrome

→ also view: Neurology p.27

CentoMito Genome

Genes: 37

CentoMito Genome includes mitochondrial genes. Nuclear genes linked to mitochondrial diseases are not included.

TAT	15 business days
Coverage	≥97.0% ≥200x
Methods	CNV analysis included ≥5.0% mitochondrial heteroplasmy can be confidently detected

Common Syndromes & Disorders Covered

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leber hereditary optic neuropathy
- Leigh-like syndrome
- Leigh syndrome
- Mitochondrial disorders
- Neurogenic muscle weakness, ataxia and retinitis pigmentosa (NARP)

→ also view: Neurology p.27

Diabetes and Obesity Panel

Genes: 265

Our Diabetes and Obesity Panel is recommended for patients with abnormalities in glucose metabolism, such as hyperinsulinemic hypoglicemia, diabetes neonatal, Maturity-onset diabetes of the young (MODY), diabetes in adults, and familial hypercholesterolemia as well as for patients displaying insulin resistance, from mild to the severe spectrum (Donohue syndrome), and for patients with familial hyperinsulinism. Prader-Willi syndrome can be detected with this panel, however, other disorders caused by imprinting errors or uniparental disomy such as 6q24-related transient neonatal diabetes mellitus and Beckwith-Wiedemann syndrome are not included in this panel.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included MLPA: 15q11

Common Syndromes & Disorders Covered

- Bardet-Biedl syndrome
- Congenital glycosylation disease
- Congenital hyperinsulinism
- Congenital hypothyroidism
- Diabetes insipidus
- Growth hormone deficiency
- Familial hypercholesterolemia
- Hypoglycemia
- Maturity-onset diabetes of the young
- Neonatal diabetes
- Obesity
- Prader-Willi syndrome

→ also view: Endocrinology p.13

Atypical Hemolytic Uremic Syndrome (aHUS) Panel

Genes: 25

Our Atypical Hemolytic Uremic Syndrome (aHUS) Panel contains genes for the molecular diagnosis of this syndrome.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included MLPA: *CFH*, *CFHR1*, *CFHR2*, *CFHR3*, *CFHR5*

Common Syndromes & Disorders Covered

- Atypical hemolytic uremic syndrome
- Methylmalonic aciduria and homocystinuria

CentoNephro

Genes: 504

Approximately 10% of the population worldwide is affected by chronic kidney diseases. Advances in genetic techniques are providing insights into kidney disease diagnosis, pathogenesis, and therapy. CentoNephro offers a comprehensive tool to diagnose the most prevalent hereditary kidney disorders, including: Alport syndrome, renal tubular acidosis, and primary hyperoxaluria, among others. CentoNephro also covers the group of disorders causing cilia dysfunction, including Joubert Syndrome, Bardet-Biedl syndrome, primary ciliary dyskinesia, Meckel syndrome, skeletal dysplasia, situs inversus, and heterotaxy syndrome, among others. *PKD1* analysis via NGS is limited due to a pseudogenic region.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included

CentoNephro Plus

Genes: 504

If polycystic kidney disease is suspected, CentoNephro Plus, which includes comprehensive analysis of *PKD1*, is recommended.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included MLPA: *PKD1* Sanger sequencing: *PKD1*

Common Syndromes & Disorders Covered

- Alport syndrome
- Bardet-Biedl syndrome
- Bartter syndrome
- Combined pituitary hormone deficiency
- Focal segmental glomerulosclerosis
- Heterotaxy syndrome
- Hypogonadotropic hypogonadism
- Intrahepatic cholestasis
- Joubert syndrome
- Kallmann syndrome
- Leber congenital amaurosis
- Meckel syndrome
- Nephronophthisis
- Nephrotic syndrome
- Neonatal mitochondrial hepatopathies
- Polycystic kidney disease*
- Pseudohypoaldosteronism
- Primary ciliary dyskinesia
- Renal tubular acidosis
- Renal tubular dysgenesis
- Skeletal dysplasia
- Skeletal ciliopathy

* Disorder only covered by CentoNephro Plus

Ataxia / Spastic Paraplegia Panel

Our Ataxia / Spastic Paraplegia Panel includes genes relevant to hereditary neurological disorders characterized by ataxia and spastic paraplegia.The covered disorders normally share overlapping symptoms and can only be clearly differentiated by molecular genetic testing. Recent information shows that ataxias and spastic paraplegia share genes, pathways and mechanisms and therefore the panel covers both syndromes and involves ataxia-spasticity disease spectrum. This is the best option for patients displaying gait imbalance and uncoordinated walking, and / or displaying spastic gait impairment, spastic weakness, and hyperreflexia or any of the combinations. The most common forms of inherited ataxia are caused by repeat expansion mutations, therefore the comprehensive version of our panel includes repeat expansion analysis.

Common Syndromes & Disorders Covered

- Cerebellar ataxia
- Episodic ataxia
- Pontocerebellar hypoplasia
- Spinocerebellar ataxia
- Spastic paraplegia, autosomal dominant
- Spastic paraplegia, autosomal recessive

Ataxia / Spastic Paraplegia Panel

Genes	483
TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included mtDNA analysis included

Ataxia Repeat Expansion Panel

Genes	13
TAT	25 business days
Methods	Repeat expansion analysis: *ATN1*, *ATXN1*, *ATXN10*, *ATXN2*, *ATXN3*, *ATXN7*, *ATXN8OS*, *BEAN1*, *CACNA1A*, *FXN*, *NOP56*, *PP2R2B*, *TBP*

Ataxia / Spastic Paraplegia Comprehensive Panel

Genes	494
TAT	25 business days
Coverage	≥ 99.0 % ≥ 20 x
Methods	CNV analysis included mtDNA analysis included Repeat expansion analysis: *ATN1*, *ATXN1*, *ATXN10*, *ATXN2*, *ATXN3*, *ATXN7*, *ATXN8OS*, *BEAN1*, *CACNA1A*, *FXN*, *NOP56*, *PP2R2B*, *TBP*

CentoICU®

Genes: 856

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TAT	15 business days FAST: 10 business days
Coverage	≥99.0% ≥20x

Common Syndromes & Disorders Covered

- Alagille syndrome
- Alpha-Thalassemia
- Arginase deficiency
- Beta-Thalassemia
- Biotinidase deficiency
- Biotin-thiamine-responsive basal ganglia disease
- Carnitine deficiency
- Congenital hypothyroidism
- Cystic Fibrosis
- Dystonia DOPA responsive
- Factor VII deficiency
- Glucose transporter 1 deficiency
- Glutaric acidemia type 1
- Hereditary fructose intolerance
- Holocarboxylase synthetase deficiency
- Maple syrup urine disease (MSUD)
- Nonketotic hyperglicinemia
- Phenylketonuria
- Pompe disease
- Primary coenzyme Q10 deficiency
- Pyridoxamine 5 phosphate oxidase deficiency
- Pyridoxine-dependent epilepsy
- Pyruvate carboxylase deficiency
- Tuberous sclerosis complex
- Tyrosinemia type I
- VLCAD deficiency

→ also view: Metabolic Disorders p.18

CentoMito Comprehensive

Genes: 451

CentoMito Comprehensive covers the entire mitochondrial genome with detection of heteroplasmy down to 15% and tests for nuclear genes related to mitochondrial diseases. Mitochondrial diseases are genetic conditions that occur when mitochondria fail to produce enough energy for the cell. Genetic mutations related to mitochondria cause symptoms mainly in organs, that consume large amounts of energy. These organs include the eye, kidney, pancreas, blood, inner ear, colon, skeletal muscle, heart, and brain.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included ≥15.0% mitochondrial heteroplasmy can be confidently detected

Common Syndromes & Disorders Covered

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leigh syndrome and maternally inherited Leigh syndrome
- Mitochondrial disorders
- Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes
- Myoclonus epilepsy with ragged red fibers
- Myogastrointestinal encephalomyopathy
- Neurogenic muscle weakness, ataxia and retinitis pigmentosa (NARP)
- Neonatal mitochondrial hepatopathies
- Pearson syndrome

→ also view: Metabolic Disorders p.21

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Genes: 37

CentoMito Genome includes mitochondrial genes. Nuclear genes linked to mitochondrial diseases are not included.

TAT	15 business days
Coverage	≥97.0% ≥200x
Methods	CNV analysis included ≥5.0% mitochondrial heteroplasmy can be confidently detected

Common Syndromes & Disorders Covered

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leber hereditary optic neuropathy
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- Leigh syndrome
- Mitochondrial disorders
- Neurogenic muscle weakness, ataxia and retinitis pigmentosa (NARP)

→ also view: Metabolic Disorders p.21

CentoNeuro

Genes: 1902

CentoNeuro is our largest panel, designed to detect an array of neurological disorders from neonatal ICU cases to dementia or movement disorders in adults. This panel includes genes related to neurological diseases, such as amyotrophic lateral sclerosis, dementia, Parkinson's disease, neuromuscular diseases, Charcot-Marie-Tooth disease, dystonia, epilepsy, autism, intellectual disability, migraine, spastic paraplegia, ataxia, Leigh syndrome, peroxisomal diseases, epileptic encephalopathies, and movement disorders, among others.

Please consider that CentoNeuro does not include repeat expansion analysis. If suspicion of neurological disorders caused by repeat expansions we recommend that physicians orders one of our disease specific panels. If there is high suspicion of Duchenne muscular dystrophy, we recommend that clinicians order deletion/duplication analysis by MLPA targeted to the *DMD* gene.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Amyotrophic lateral sclerosis
- Arthrogryposis multiplex congenita
- Ataxia
- Dementia
- Dolichoectasia
- Dystonia
- Epilepsy
- Familial hemiplegic migraine
- Frontotemporal dementia
- Hypogonadotropic hypogonadism
- Intellectual disability
- Joubert syndrome
- Kallman syndrome
- Leigh syndrome
- Leukodystrophy and peroxisome biogenesis disorders
- Meckel syndrome
- Mitochondrial encephalomyopathy
- Neonatal mitochondrial hepatopathies
- Neuromuscular disorders
- Parkinson's disease
- Refsum disease
- Spastic paraplegia
- Tuberous sclerosis
- Zellweger syndrome

Amyotrophic Lateral Sclerosis (ALS) / Dementia Panel

Genes: 105

Our Amyotrophic Lateral Sclerosis (ALS) / Dementia Panel is designed to detect ALS, which is a progressive neurodegenerative disorder characterized by the degeneration of the upper and lower motor neurons. In addition, our panel includes genes causing Alzheimer disease, dementia, and frontotemporal dementia, as well as to differentially diagnose among diseases with overlapping symptoms. Genes included in this panel have been carefully selected to increase the diagnostic yield. Actionable diseases overlapping with the phenotype are included such as Wilson disease, Niemann-Pick disease, and hexosaminidase A deficiency. This panel does not detect Huntington disease.

TAT	25 business days
Coverage	≥ 99.0% ≥ 20 x
Methods	CNV analysis included mtDNA analysis included Repeat expansion analysis: *ATXN2*, *C9orf72*, *PRNP*

Common Syndromes & Disorders Covered

- Alzheimer disease
- Amyotrophic lateral sclerosis
- Dementia
- Frontotemporal dementia
- Hexosaminidase A deficiency
- Niemann-Pick disease
- Wilson disease

Epilepsy Panel

Genes: 783

While some types of seizures are easily categorized (i.e., partial or generalized), others are not or might later develop into different types, like partial seizures with secondary generalization, making targeted panel testing less likely to succeed at reaching a diagnosis. Our Epilepsy Panel is phenotype-directed and covers different types of seizure syndromes, covering Dravet syndrome, early infantile epileptic encephalopathy, epilepsy partial, epilepsy generalized, epilepsy absence, myoclonic epilepsy, and hypomagnesemia. In addition, our panel includes mitochondrial and nuclear mitochondrial genes, including those associated with myoclonic epilepsy with ragged red fibers (MERRF).

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included Repeat expansion analysis: *CSTB*

Common Syndromes & Disorders Covered

- Aicardi-Goutieres syndrome
- Brain iron accumulation syndromes
- Congenital glycosylation disease
- Dravet syndrome
- Early infantile epileptic encephalopathy
- Epilepsy
- Epilepsy (absence) in childhood
- Epilepsy (generalized) with febrile seizures
- Epilepsy (partial)
- Epileptic encephalopathy
- Hypomagnesemia
- Leigh syndrome
- Leukodystrophy and peroxisome biogenesis disorders
- Lysosomal storage disease
- Mitochondrial DNA depletion
- Mitochondrial encephalomyopathy
- Muscular dystrophy-dystroglycanopathy
- Myoclonic epilepsy
- Urea cycle disorder

Intellectual Disability Panel

Genes: 820

Our panel includes genes associated with intellectual disabilities covering all mechanisms of inheritance as well as syndromic and non-syndromic autism, microcephaly, neuronal migration disorders, developmental regression, and Aicardi-Goutieres syndrome. Detection of fragile X syndrome is possible: our panel includes the detection of repeat expansion of *FMR1*.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included Repeat expansion analysis: *FMR1*

Common Syndromes & Disorders Covered

- Aicardi-Goutieres syndrome
- Bardet-Biedl syndrome
- Epileptic encephalopathy
- Intellectual disability AD, AR, XL
- Micro syndrome
- Microcephaly
- Neurodevelopmental disorders
- Neuronal migration disorders
- Syndromic autism

Neuromuscular Panel

Genes: 355

Our Neuromuscular Panel is ideal for patients with muscular diseases. It includes genes causing neurological diseases and covers disorders, such as metabolic myopathies, muscular dystrophies, Charcot-Marie-Tooth disease, congenital myasthenic syndrome, congenital myopathies, myofibrillar myopathies, nemaline myopathies, and other syndromes with hypotonia, myotonia or weakness. Arthrogryposis is included for differential diagnosis of early-onset neuromuscular disorders. If there is high diagnostic suspicion of Duchenne muscular dystrophy, we recommend that clinicians order deletion/duplication analysis by MLPA targeted to the *DMD* gene as an additional service.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included MLPA: *SMN1* and *SMN2* Repeat Expansion: *DMPK*

Common Syndromes & Disorders Covered

- Arthrogryposis
- Bethlem myopathy
- Charcot-Marie-Tooth disease
- Congenital myasthenic syndrome
- Congenital myopathy
- Dejerine-Sottas syndrome
- Hyperekplexia
- Hypotonia
- Malignant hyperthermia
- Metabolic myopathies
- Muscular dystrophy
- Muscular dystrophy-dystroglycanopathy type A
- Myofibrillar myopathy
- Myopathy-rhabdomyolysis syndrome
- Nemaline myopathy
- Non-dystrophic myotonia congenita
- Spinal muscular atrophy type 1
- Ullrich muscular dystrophy

Parkinson's Disease Panel

Genes: 115

Our Parkinson's Disease (PD) Panel identifies all relevant pathophysiologically genetic variants linked to the development and treatment of PD. Characteristic features of PD include neuronal loss in specific areas of the substantia nigra and widespread intracellular protein α-synuclein accumulation. The disease is characterized by three core motor symptoms: tremor, muscle rigidity, and bradykinesia.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Alzheimer disease
- Basal ganglia calcification
- Niemann-Pick disease
- Parkinson's disease
- Striatal degeneration

BRCA1, *BRCA2*

Genes: 2

Mutations in *BRCA1* and *BRCA2* can increase the risk of developing cancer. Mutations in these two genes are responsible for 5–10 % of all breast cancers in women.

Common Syndromes & Disorders Covered

- Breast cancer

BRCA1, *BRCA2* Plus

TAT	15 business days
Coverage	≥ 99.0 % ≥ 20 x
Type	Germline
Methods	CNV analysis included

BRCA1, *BRCA2* Combi

TAT	15 business days
Coverage	≥ 99.0 % ≥ 20 x
Type	Germline
Methods	MLPA included

BRCA1, *BRCA2* Somatic

TAT	10 business days
Coverage	variable
Type	Somatic

CentoBreast

Genes: 28

CentoBreast detects mutations in the *BRCA1* and *BRCA2* genes, which are the most common hereditary causes of breast cancer. In addition, our panel includes other genes such as *ATM*, *BRIP1*, *CHEK2*, *PALB2*, etc. which have also been associated with increased cancer risk.

Breast cancer is one of the most common cancers in the world affecting ~12.5% of women during their lifetime, with 5–10% of these patients having a hereditary form.

TAT	15 business days
Coverage	≥99.0% ≥20x
Type	Germline
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Breast cancer
- Ovarian cancer

CentoCancer®

Genes: 67

Each gene in CentoCancer® has been carefully selected based on its potential to contribute to the risk of developing one or more of the following cancers: breast cancer, ovarian cancer, colorectal cancer, gastric cancer, thyroid cancer, endometrial cancer, pancreatic cancer, melanoma, renal cancer, and prostate cancer. This panel is appropiate for patients with a family history of early-onset cancer, rare cancer, bilateral cancer, or multiple primary cancers.

TAT	15 business days
Coverage	≥99.0% ≥20x
Type	Germline
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Breast cancer
- Colorectal cancer
- Endometrial cancer
- Familial adenomatous polyposis
- Gastric cancer
- Gastrointestinal stromal tumor
- Melanoma
- Ovarian cancer
- Pancreatic cancer
- Prostate cancer
- Renal cancer
- Skin cancer
- Thyroid cancer
- Uterine cancer

CentoCancer® Comprehensive

Genes: 118

CentoCancer® Comprehensive is our most extensive cancer panel, covering a large number of cancer-associated genes. Each gene in this panel has been carefully selected based on its potential to contribute to the risk of developing one or more of the following cancers: breast cancer, ovarian cancer, colorectal cancer, gastric cancer, thyroid cancer, endometrial cancer, pancreatic cancer, melanoma, renal cancer, prostate cancer, among others.

TAT	15 business days
Coverage	≥99.0% ≥20x
Type	Germline
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Breast cancer
- Colorectal cancer
- Endometrial cancer
- Familial adenomatous polyposis
- Gastric cancer
- Gastrointestinal stromal tumor
- Hereditary paraganglioma / pheochromocytoama
- Melanoma
- Ovarian cancer
- Pancreatic cancer
- Paragangliomas / pheochromocytoma / gastrointestinal stromal
- Prostate cancer
- Renal cancer
- Retinoblastoma
- Skin cancer
- Thyroid cancer
- Uterine cancer

CentoCancer® Pediatric

Genes: 98

Cancer occurs in people of all ages, with some being nearly exclusively tied to childhood. CentoCancer® Pediatric is our comprehensive solution to detect genes associated with pediatric cancer. The gene list has been carefully curated by internal and external experts to cover the most common forms of pediatric cancer. Germline mutations identified by these panel will help to define prognosis, differentiate patient / family risk, and guide treatment decisions. Spotting cancer early increases the chances of survival.

TAT	15 business days
Coverage	≥ 99.0% ≥ 20 x
Type	Germline
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Bone cancer
- Leukemia
- Lymphomas
- Malignant brain tumors
- Neuroblastoma
- Rhabdomyosarcoma
- Wilms tumor

Myeloid Tumor Panel

Genes: 35

Our Myeloid Tumor Panel targets important regions within 35 genes that are frequently mutated in myeloid malignancies. Myeloid malignancies are clonal diseases of hematopoietic progenitor cells. Myeloid tumors represent the fourth most frequently diagnosed cancer in economically developed countries. The majority of myeloid tumors contain high numbers of somatic mutations, which are genetic changes that are not inherited but created within the tumor itself. Unlike inherited "germline" mutations, these somatic mutations are not transmitted to offspring. Somatic mutations significantly contribute to the pathogenesis, progression, and prognosis of myeloid malignancies.

TAT	10 business days
Coverage	≥97.0% ≥200x
Type	Somatic

Common Syndromes & Disorders Covered

- Acute myeloid leukemia
- Chronic myeloid leukemia
- Chronic myelomonocytic leukemia
- Juvenile myelomonocytic leukemia
- Myelodysplastic syndrome
- Myeloid tumor
- Myeloproliferative neoplasms

Solid Tumor Panel

Genes: 149

Our Solid Tumor Panel provides full sequencing of 106 selected cancer-associated genes and the hotspot analysis of relevant cancer regions in 43 genes. It detects over 5,000 validated oncogenic variants and includes the latest evidence-based variants associated with treatment decisions in solid tumors. The panel includes more than 25 genes associated with approved targeted therapies or treatments that are currently being tested in clinical trials. Furthermore, somatic variants with an impact on prognosis of the individual tumor or the efficacy of standard anti-tumor therapy are captured. It covers more than 100 different types of somatic cancers, including adrenal cancer, colon cancer, hepatic cancer, prostate cancer, renal cancer, skin cancer, testicular cancer, thyroid cancer, glioma, esophageal cancer, endometrial cancer, and breast cancer, among others. The panel provides a better understanding of tumor behavior as well as its likelihood to respond to a treatment – thus frequently leading to better outcomes or reduced adverse effects.

TAT	10 business days
Coverage	≥97.0% ≥200x
Type	Somatic

Common Syndromes & Disorders Covered

- Adrenal cancer
- Biliary tract cancer
- Bone marrow cancer
- Breast cancer
- Colon cancer
- Endometrial cancer
- Esophageal cancer
- Gastrointestinal stromal tumor
- Glioma
- Hepatic cancer
- Lung cancer
- Lymphoma cancer
- Meningioma
- Ovarian cancer
- Pancreatic cancer
- Prostate cancer
- Renal cancer
- Skin cancer
- Testicular cancer
- Thyroid cancer

CentoVision

Genes: 450

CentoVision is carefully designed to find the genetic basis of eye diseases, including those that are the leading causes of blindness among infants (Leber congenital amaurosis), children (early-onset retinitis pigmentosa), and adults (pattern dystrophy). Our panel includes the most common ophthalmology diseases, such as congenital glaucoma, retinitis pigmentosa, Stargardt disease, Stickler syndrome, achromatopsia, and Usher syndrome, among others. It also screens for different types of albinism (oculocutaneous and ocular) as well as Hermasky-Pudlak syndrome.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Achromatopsia
- Albinism
- Bardet-Biedl syndrome
- Cataract
- Cone-rod and cone dystrophy
- Flecked retina
- Glaucoma
- Hermansky-Pudlak syndrome
- Leber congenital amaurosis
- Meckel syndrome
- Microphthalmia / anophthalmia / coloboma spectrum
- Oculomotor apraxia
- Optic atrophy
- Progressive external ophthalmoplegia
- Retinitis pigmentosa, autosomal dominant
- Retinitis pigmentosa, autosomal recessive
- Stargardt disease
- Stickler syndrome
- Usher syndrome
- Vitreoretinopathy
- Wagner syndrome

Abnormal Mineralization Panel

Genes: 94

Our Abnormal Mineralization Panel includes detection of genes causing osteogenesis imperfecta, osteopetrosis, high and low bone density disorders, and genes necessary to discriminate a true genetic cause. Diseases with medical management options, such as hypophosphatasia, are also included in this panel.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included

Common Syndromes & Disorders Covered

- Abnormal mineralization
- High bone density disorders
- Low bone density disorders
- Osteogenesis imperfecta
- Osteopetrosis

Pulmonary Panel

Genes: 101

Our Pulmonary Panel includes genes to diagnose central hypoventilation syndrome, surfactant metabolism dysfunction, and pulmonary hypertension, among other pulmonary diseases.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included Repeat expansion analysis: *PHOX2B*

Common Syndromes & Disorders Covered

- Central hypoventilation syndrome
- Comprehensive pulmonary disease
- Pulmonary hypertension
- Surfactant metabolism dysfunction

CentoScreen®

Genes: 330

CentoScreen® is our comprehensive carrier screening panel, covering more than 300 common autosomal recessive and X-linked disorders. It provides the opportunity to make informed decisions and review the range of options available to guide pregnancy and family planning.

CentoScreen® Solo

TAT	25 business days
Coverage	≥99.0 % ≥20 x
Methods	CNV analysis* included MLPA: *SMN1* Repeat expansion analysis: *FMR1* Sanger sequencing: *CYP21A2*

CentoScreen® Duo

TAT	25 business days
Coverage	≥99.0 % ≥20 x
Details	CNV analysis* for each partner included MLPA: *SMN1* Repeat expansion analysis: *FMR1* Sanger sequencing: *CYP21A2*

CentoScreen® Paired

Includes complete panel evaluation with CNV analysis* of 34 genes + risk gene analysis of partner.

TAT	30 business days
Coverage	≥99.0 % ≥20 x
Details	MLPA: *SMN1*** Repeat expansion analysis: *FMR1*** Sanger sequencing: *CYP21A2***

Common Syndromes & Disorders Covered

- Alport disease
- Bardet-Biedl syndromes
- Congenital adrenal hyperplasia
- Cystic fibrosis
- Fragile X syndrome
- Glycine encephalopathy
- Maple syrup urine disease
- Mucolipidosis, several types
- Mucopolysaccharidosis, several types
- Niemann-Pick disease
- Spinal muscular atrophy
- Organic acidemias
- Wilson disease

* CNV analysis included for: *ABCC6*, *ALDH3A2*, *COL4A5*, *CTNS*, *DBT*, *DMD*, *EDA*, *F8*, *FANCA*, *FKTN*, *GAA*, *GALC*, *GBE1*, *GJB6*, *GLDC*, *HBA1*, *HBA2*, *HBB*, *HEXB*, *HPRT1*, *HPS3*, *HSD17B4*, *IDS*, *MCOLN1*, *NEB*, *OTC*, *PAH*, *PCCA*, *PCDH15*, *PDHA1*, *RAPSN*, *SGCB*, *STS* and *XPC*

** only for index patient

Fertility Panel

Genes: 276

Our Fertility Panel is recommended for couples trying to conceive for one year or longer, who have experienced more than one miscarriage, with irregular or absent menstruation, with low sperm count, form, or movement, or with absence of development of secondary sexual features. Knowing the exact cause of infertility allows for better clinical management and enables enhanced counseling and personalized reproductive care plan for couples. This NGS-only panel option does not include sex chromosome aneuploidy, Y chromosome microdeletions and *FMR1*-related premature ovarian failure syndrome testing. For a more comprehensive analysis, please refer to our Fertility X and Fertility Y panels.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included

Common Syndromes & Disorders Covered

- Androgen insensitivity syndrome
- Hypogonadotropic hypogonadism
- Monogenic causes of female infertility
- Monogenic causes of male infertility
- Ovarian hyperstimulation syndrome
- Premature ovarian failure (not *FMR1*-related)
- Primary ciliary dyskinesia
- Spermatogenic failure

Fertility X Panel

Genes: 276

Our Fertility X Panel is recommended for patients assigned female gender at birth trying to conceive for one year or longer, who have experienced more than one miscarriage, with irregular or absent menstruation, or with absence of development of secondary sexual features. Our panel includes the most important genes related to infertility in females. Knowing the exact cause of infertility allows for better clinical management and enables enhanced counseling and personalized reproductive care plan for couples.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included Repeat expansion analysis: *FMR1* MLPA: Aneuploidy

Common Syndromes & Disorders Covered

- Complete androgen insensitivity syndrome
- Hypogonadotropic hypogonadism
- Monogenic causes of female infertility
- Ovarian hyperstimulation syndrome
- Premature ovarian failure
- Turner syndrome

Fertility Y Panel

Genes: 276

Our Fertility Y Panel is recommended for patients assigned male gender at birth with known fertility problems, part of a couple that has experienced more than one miscarriage, with low sperm count, form, or movement, or with absence of development of secondary sexual features. Our panel includes the most important genes related to infertility in males. Knowing the exact cause of infertility allows for better clinical management and enables enhanced counseling and personalized reproductive care plan for couples.

TAT	25 business days
Coverage	≥99.0% ≥20x
Methods	CNV analysis included mtDNA analysis included MLPA: Aneuploidy, AZF region

Common Syndromes & Disorders Covered

- CFTR-associated infertility
- Hypogonadotropic hypogonadism
- Klinefelter syndrome
- Monogenic causes of male infertility
- Primary ciliary dyskinesia
- Spermatogenic failure
- Y chromosome microdeletions

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