CENTOGENE
THE RARE DISEASE COMPANY

NGS Panels

A Targeted Approach for Testing Genetic Disorders

Working together…

NGS Panels

BENEFIT FROM OUR MEDICAL EXPERTISE AND STREAMLINED GENETIC TESTING

CENTOGENE is fully committed to bringing the best possible diagnostic solutions to our 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. To reflect the fast-growing knowledge of complex associations of gene-disease associations and to maximize clinical sensitivity, we have updated and significantly redesigned our Next Generation Sequencing (NGS) gene panels.

The gene composition of each panel has been revised to include the latest gene discoveries and to provide the highest clinical certainty. Additionally, we have minimized complexity and removed redundancy in the panel portfolio by creating comprehensive phenotype-directed diagnostic panels. They include all relevant genes necessary for differential diagnosis of syndromes with overlapping phenotype — allowing diseases that otherwise would be missed to be diagnosed. 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 — significantly simplifying the diagnostic process for you and your patients. As always, CENTOGENE's Customer Support Team is available to answer questions and help in the diagnostic process in any way we can.

Content

Panel Specifications

CENTOGENE PANEL	≥99.0% targeted regions covered at ≥20x. For each gene, all single nucleotide variants described in HGMD® and the CENTOGENE Biodatabank are covered, including relevant 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).
COMPLEMENTARY ASSAYS	Some panels can be 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/ngs-panels
DELETION/DUPLICATION	NGS-based copy number variations (CNV) are detected with a sensitivity of above 95.0% 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

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(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; lack of sufficient clinical information; and in our oncogenetic panels.
REQUESTED MATERIAL	1 CentoCard®*
TAT	25 days

* Except for: *BRCA1, BRCA2* somatic and solid tumor panel, where the requested material is FFPE tissue (block or sections) or fresh tumor tissue. For more details of accepted materials please check: centogene.com/how-to-order

Disclaimer:
Due to continous developments in our product portfolio the gene numbers in our panels are subject to change without prior notice.
List 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

CENTOGENE PANEL

CentoCardio

Genes: 327

CentoCardio includes the most relevant genes for arrythmias, congenital heart disease, and cardiomyopathies. Syndromes included: Long and short QT, 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.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
mtDNA analysis included

COMMON SYNDROMES AND 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

CENTOGENE PANEL

CentoSkin

Genes: 152

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 section.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included

COMMON SYNDROMES AND 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

CENTOGENE PANEL

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, Costello syndrome, Cardio-Facio-Cutaneous syndrome, and Legius syndrome, among others. Tuberous sclerosis and McCune Albright syndrome are included for differential diagnosis.

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included
mtDNA analysis included

COMMON SYNDROMES AND 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

CENTOGENE PANEL

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, cutis laxa, Ehlers-Danlos, Stickler syndrome, and familial thoracic aortic aneurysm and dissection.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

CentoHear

Genes: 196

Hearing loss is a common condition in children, affecting 1 in 100 live births. In more than 50.0% of cases, there is a genetic cause for this disorder, of which 70.0% 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, Pendred, Waardenburg, Usher, and branchio-oto-renal, among others.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included

COMMON SYNDROMES AND 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

CENTOGENE PANEL

COMMON SYNDROMES AND DISORDERS COVERED

Diabetes and obesity panel

Genes: 265

Our diabetes and obesity panel is recommended for patients with abnormalities in glucose metabolism, such as hyperinsulinemic hypoglycemia, diabetes neonatal, 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. Disorders caused by imprinting errors or uniparental disomy such as 6q24-related transient neonatal diabetes mellitus and Beckwith Wiedemann syndrome are not detected with this panel.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
MLPA: 15q11

- 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

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.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included

- Pancreatic cancer
- Pancreatitis

CENTOGENE PANEL

Congenital adrenal hyperplasia 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 90.0% of CAH cases are caused by a deficiency of this enzyme.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
MLPA: *CYP21A2*
Sanger sequencing: *CYP21A2*

COMMON SYNDROMES AND DISORDERS COVERED

- Congenital adrenal hyperplasia
- Pigmented nodular adrenocortical disease

CENTOGENE PANEL

Blood coagulation panel

Genes: 112

Our blood coagulation panel contains genes to diagnose thrombophilia, thrombocytopenia, hereditary hemorrhagic telangiectasia, ARC syndrome, Hermasky-Pudlak syndrome, coagulation factor disorders, hemophilia, and platelet related disorders.

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

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

Bone marrow failure / Anemia panel

Genes: 212

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, and dyserythropoietic anemia.

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included

COMMON SYNDROMES AND 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
- Sitosterolemia
- Thrombocytopenia

CENTOGENE PANEL

CentoImmuno

Genes: 330

CentoImmuno is our solution for immunodeficiency and severe combined immunodeficiency (SCID) disorders. Our panel includes genes targeting severe combined immunodeficiency, congenital neutropenia, primary antibody deficiency, common variable immune deficiency, chronic granulomatous disease, autoimmune lymphoproliferative, afibrinogenemia, hemolytic uremic syndrome, and agammaglobulinemia.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included

COMMON SYNDROMES AND DISORDERS COVERED

- Agammaglobulinemia
- Autoimmune lymphoproliferative syndrome
- B-cell-negative severe combined immunodeficiency
- B-cell-positive severe combined immunodeficiency
- Bare lymphocyte syndrome
- Chronic granulomatous disease
- Common variable immune deficiency
- Complement deficiency
- Congenital afibrinogenemia
- Congenital neutropenia syndromes
- Hermasky-Pudlak syndrome
- Hemolytic uremic syndrome
- Mendelian susceptibility to mycobacterial diseases
- Periodic fever syndrome
- Primary antibody deficiency
- Primary ciliary dyskinesia
- Primary immunodeficiencies (PID)
- Psoriasis
- Severe combined immunodeficiency

CENTOGENE PANEL

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.

15 days TAT; ≥ 99.0 % ≥ 20 x coverage
FAST option: 10 days TAT

COMMON SYNDROMES AND 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)
- Non ketotic 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

CENTOGENE PANEL

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.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
mtDNA analysis included

COMMON SYNDROMES AND 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
- Non-ketotic hyperglycinemia
- Organic acidemias
- Porphyria
- Refsum disease
- Urea cycle disorder

CENTOGENE PANEL

CentoMetabolic MOx

Genes: 206

CentoMetabolic® 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.

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

COMMON SYNDROMES AND 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

CENTOGENE PANEL

COMMON SYNDROMES AND DISORDERS COVERED

CentoMito comprehensive

Genes: 451

CentoMito comprehensive covers the entire mitochondrial genome with detection of heteroplasmy down to 15.0% 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.

25 days TAT; ≥99.0% ≥20x coverage
≥15.0% mitochondrial heteroplasmy can be confidently detected
CNV analysis included

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leigh's syndrome and maternally inherited Leigh's syndrome
- Mitochondrial disorders
- Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes
- Myoclonus epilepsy with ragged red fibers
- Myogastrointestinal encephalomyopathy
- NARP
- Neonatal mitochondrial hepatopathies
- Pearson syndrome

CentoMito Genome

Genes: 37

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

25 days TAT; ≥97.0% ≥200x coverage
≥15.0% mitochondrial heteroplasmy can be confidently detected
CNV analysis included

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leber hereditary optic neuropathy
- Leigh-like syndrome
- Leigh syndrome
- Mitochondrial disorders
- NARP

CENTOGENE PANEL

Diabetes and obesity panel

Genes: 265

Our diabetes and obesity panel is recommended for patients with abnormalities in glucose metabolism, such as hyperinsulinemic hypoglycemia, diabetes neonatal, 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. Disorders caused by imprinting errors or uniparental disomy such as 6q24-related transient neonatal diabetes mellitus and Beckwith Wiedemann syndrome are not detected with this panel.

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included
MLPA: 15q11

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

Atypical hemolytic uremic syndrome panel

Genes: 25

Our atypical hemolytic uremic syndrome panel contains genes for the molecular diagnosis of this syndrome.

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

COMMON SYNDROMES AND DISORDERS COVERED

- Atypical hemolytic uremic syndrome
- Methylmalonic aciduria and homocystinuria

CENTOGENE PANEL

CentoNephro

Genes: 502

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 screen for the most prevalent hereditary kidney disorders, including: alport syndrome, renal tubular acidosis panel, focal glomerulonephrosis panel, and primary hyperoxaluria, among others. CentoNephro also covers the group of disorders causing cilia dysfunction, including Joubert Syndrome, Bardet-Biedl, COACH syndrome, primary ciliary dyskinesia, Meckel syndrome, skeletal dysplasia, situs inversus, and heterotaxy, among others. *PKD1* analysis is not included in this panel.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included

CentoNephro Plus

Genes: 503

If polycystic kidney disease is suspected, CentoNephro Plus is recommended, which includes all genes from CentoNephro and *PKD1* analysis.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
MLPA: *PKD1*
Sanger sequencing: *PKD1*

* Disorder only covered by CentoNeprho Plus

COMMON SYNDROMES AND 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

CENTOGENE PANEL

Ataxia / Spastic paraplegia panel

Our Ataxia / Spastic paraplegia panel includes genes relevant to hereditary neurological disorders characterized by ataxia and spastic paraplegia, including spinocerebellar ataxia (dominant and recessive), cerebellar ataxia, episodic ataxia, and pontocerebellar ataxia. These disorders normally share overlapping symptoms and can only be clearly differentiated by molecular genetic testing. Traditionally, ataxias and spastic paraplegia have been classified into separate categories. However, recent information shows that these diseases share genes, pathways and mechanisms and therefore our panel covers both syndromes and involves ataxia-spasticity disease spectrum. Our Ataxia / Spastic paraplegia panel is not only the best option for patients displaying gait imbalance and unco-ordinated walking, but also for patients 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 compre-hensive version of our panel includes repeat expansion analysis.

.Ataxia / Spastic paraplegia panel

Genes: 482
25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included
mtDNA analysis included

Ataxia / Spastic paraplegia comprehensive panel

Genes: 493
25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included
mtDNA analysis included
Repeat expansion analysis: *ATN1*, *ATXN1*, *ATXN10*, *ATXN2*, *ATXN3*, *ATXN7*, *ATXN8OS*, *BEAN1*, *CACNA1A*, *FXN*, *NOP56*, *PP2R2B*, *TBP*

COMMON SYNDROMES AND DISORDERS COVERED

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

Ataxia repeat expansion panel

Genes: 13
Includes repeat expansion analysis;
25 days TAT; 100 % coverage
Repeat expansion analysis: *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS*, *BEAN1*, *CACNA1A*, *FXN*, *NOP56*, *PP2R2B*, *TBP*

CENTOGENE PANEL

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.

15 days TAT; ≥99.0% ≥20x coverage
FAST option: 10 days TAT

COMMON SYNDROMES AND 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)
- Non ketotic 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

CENTOGENE PANEL

COMMON SYNDROMES AND DISORDERS COVERED

CentoMito comprehensive

Genes: 451

CentoMito comprehensive covers the entire mitochondrial genome with detection of heteroplasmy down to 15.0% 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.

≥ 15.0 % mitochondrial heteroplasmy can be confidently detected
CNV analysis included

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leigh's syndrome and maternally inherited Leigh's syndrome
- Mitochondrial disorders
- Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes
- Myoclonus epilepsy with ragged red fibers
- Myogastrointestinal encephalomyopathy
- NARP
- Neonatal mitochondrial hepatopathies
- Pearson syndrome

CentoMito Genome

Genes: 37

CentoMito Genome includes mitochondrial genes. Nuclear mitochondrial genes are not included.

25 days TAT; ≥97.0% ≥200x coverage
≥5.0% mitochondrial heteroplasmy can be confidently detected
CNV analysis included

- Chronic progressive external ophthalmoplegia
- Kearns-Sayre syndrome
- Leber hereditary optic neuropathy
- Leigh-like syndrome
- Leigh syndrome
- Mitochondrial disorders
- NARP

CENTOGENE PANEL

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, neuromuscular diseases, Charcot-Marie-Tooth, 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.

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included

COMMON SYNDROMES AND 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

CENTOGENE PANEL

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's, 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, and hexosaminidase A deficiency. This panel does not detect Huntington's disease.

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included
mtDNA analysis included
Repeat expansion analysis: *ATXN2*, *C9ORF72*, *PRNP*

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

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 — i.e., 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 panel, and hypomagnesemia. In addition, our panel includes mitochondrial and nuclear mitochondrial genes (i.e., genes causing myoclonic epilepsy with ragged red fibers –MERRF–).

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
mtDNA analysis included
Repeat expansion analysis: *CSTB*

COMMON SYNDROMES AND 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

CENTOGENE PANEL

Intellectual disability panel

Genes: 819

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 Goutierres. Detection of Fragile X syndrome is possible: our panel includes the detection of repeat expansion of *FMR1*.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
mtDNA analysis included
Repeat expansion analysis: *FMR1*

COMMON SYNDROMES AND 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

CENTOGENE PANEL

Neuromuscular panel

Genes: 354

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, 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.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
E7 homozygous deletion screening: *SMN1*
mtDNA analysis included
Repeat Expansion: *DMPK*

COMMON SYNDROMES AND DISORDERS COVERED

- Arthrogryposis
- Bethlem myopathy
- Charcot–Marie–Tooth disease
- Congenital myasthenic syndrome
- Congenital myopathy
- Dejerine-Sottas syndrome
- E7 homozygous deletion screening: *SMN1*
- 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

CENTOGENE PANEL

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.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
mtDNA analysis included

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL	COMMON SYNDROMES AND DISORDERS COVERED
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 to 10% of all breast cancers in women. **BRCA1, BRCA2** Panel includes NGS 15 days TAT; ≥99,5% ≥20x coverage; Type: Germline **BRCA1, BRCA2 Plus** Panel includes NGS and CNV analysis 15 days TAT; ≥99,5% ≥20x coverage; Type: Germline **BRCA1, BRCA2 Combi** Panel includes NGS and MLPA 15 days TAT; ≥99,5% ≥20x coverage; Type: Germline **BRCA1, BRCA2 Somatic** Panel includes NGS 10 days TAT; variable coverage; Type: Somatic	• Breast cancer
CentoBreast Genes: 30 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, RAD51*, 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. 15 days TAT; ≥99.5% ≥20x coverage CNV analysis inluded Type: Germline	• Breast cancer • Ovarian cancer

CENTOGENE PANEL

CentoCancer®

Genes: 70

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, ovarian, colorectal, gastric, thyroid, endometrial, pancreatic, melanoma, renal, and prostate. This panel is appropiate for patients with a family history of early-onset cancer, rare cancer, bilateral cancer, or multiple primary cancers.

15 days TAT; ≥99.5% ≥20x coverage
CNV analysis inluded
Type: Germline

COMMON SYNDROMES AND 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

CENTOGENE PANEL

CentoCancer® comprehensive

Genes: 110

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, ovarian, colorectal, gastric, thyroid, endometrial, pancreatic, melanoma, renal, prostate, among others.

15 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
Type: Germline

COMMON SYNDROMES AND DISORDERS COVERED

- Beckwith-Wiedemann syndrome
- 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
- Rothmund-Thomson syndrome (Type 2)
- Skin cancer
- Thyroid cancer
- Uterine cancer

CENTOGENE PANEL

CentoColon

Genes: 33

CentoColon detects genes that are associated with colon, pancreatic, and gastric cancer.

15 days TAT; ≥99.5% ≥20x coverage
CNV analysis included
Type: Germline

COMMON SYNDROMES AND DISORDERS COVERED

- Colorectal cancer
- Familial adenomatous polyposis
- Gastric cancer
- Hereditary nonpolyposis colorectal cancer
- Pancreatic cancer

CENTOGENE PANEL

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. Diseases covered in this panel include: Acute myeloid leukemia (AML), chronic myeloid leukemia (CML), myelodysplastic syndrome (MDS), myeloproliferative neoplasms (MPN), chronic myelomonocytic leukemia (CMML), and juvenile myelomonocytic leukemia (JMML).

10 days TAT; >97.0 % >200 x coverage
Type: Somatic

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

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, colon, hepatic, prostate, renal, skin, testicular, thyroid, glioma, esophageal, endometrial, 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.

10 days TAT; >97.0 % >200 x coverage
Type: Somatic

COMMON SYNDROMES AND 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

CENTOGENE PANEL

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.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
mtDNA analysis included

COMMON SYNDROMES AND 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

CENTOGENE PANEL

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 our panel.

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

Pulmonary panel

Genes: 101

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

25 days TAT; ≥ 99.0 % ≥ 20 x coverage
CNV analysis included
Repeat expansion analysis: *PHOX2B*

COMMON SYNDROMES AND DISORDERS COVERED

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

CENTOGENE PANEL

CentoScreen®

Genes: 330

CentoScreen® is our comprehensive screening panel that covers 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

Includes complete panel evaluation with CNV analysis* of 34 genes

25 days TAT; ≥99.0% ≥20x coverage
MLPA: *SMN1*
Repeat expansion analysis: *FMR1*
Sanger sequencing: *CYP21A2*

CentoScreen® Duo

Includes complete panel evaluation with CNV analysis* of 34 genes for each partner

25 days TAT; ≥99.0% ≥20x coverage

CentoScreen® Paired

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

30 days TAT; ≥99.0% ≥20x coverage
Repeat expansion analysis for *FMR1*; and MLPA for *SMN1* are offered only for the first patient. To second patient we only offer sequencing and CNV analysis
Sanger sequencing for *CYP21A2*

COMMON SYNDROMES AND 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*

CENTOGENE PANEL

Infertility panel

Genes: 276

Our infertility panel is recommended for patients with the following fertility issues: unsuccessfully trying to conceive for longer than one year, known infertility, more than one miscarriage, irregular or absent menstruation; male partners with low sperm count, irregularities in sperm form or movement. It is also recommended for patients who fail to develop secondary sexual features. Understanding the cause of infertility enables counseling on treatment options.

25 days TAT; ≥99.0% ≥20x coverage
CNV analysis included
MLPA: Aneuploidy, AZF region
mtDNA analyisis included
Repeat expansion analysis: *AR*, *FMR1*

COMMON SYNDROMES AND DISORDERS COVERED

- Androgen insensitivity syndrome
- Female infertility
- Germline aneuploidy of chromosomes 13, 18, 21 and X
- Hypogonadotropic hypogonadism
- Klinefelter syndrome
- Male infertility
- Ovarian hyperstimulation syndrome
- Premature ovarian failure
- Primary ciliary dyskinesia
- Spermatogenic failure
- Turner Syndrome
- Y chromosome microdeletions

The CENTOGENE Advantage

MORE THAN STREAMLINED GENETIC TESTING. THE SUPPORT YOU NEED TODAY.

CentoCard®

Our quick, cost-effective, and hassle-free solution for shipment of clinical blood samples for genetic testing. CentoCard® provides a single sample for complete patient diagnostics: enzyme assay, biomarker analysis, and genetic 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.

Multiomics 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®

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

The CENTOGENE Biodatabank

The world's largest real-world data repository for rare and neurodegenerative diseases

Clinical Studies and Pharma Partnerships

By participating in clinical studies, patients benefit by contributing to the development of new therapies and improved disease monitoring. Through pharmaceutical partnerships, we also leverage our expertise to speed up drug development in rare diseases.

World-Class Expertise

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

... for a patients' better tomorrow.

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