

MOLECULAR DIAGNOSIS OF A GENETIC DISEASE BY NGS
EXOME AND GENE PANELS

SAMPLING

Sampling date:

Customer:

PRENATAL DIAGNOSIS: please include a 5mL maternal EDTA total blood sample so that we can test for maternal contamination:

- ☐ Amniotic liquid (FRESH) ☐ Amniotic liquid (CULTURE) ☐ Extracted fetal DNA
☐ Chorionic villusitis ☐ Chorionic villusitis (CULTURE) ☐ Fetal blood

POST-NATAL DIAGNOSIS:

☐ EDTA total blood (0.5mL to 5mL)

FETOPATHOLOGY:

☐ Fetal biopsy ☐ Extracted DNA

PATIENT

SURNAME
FIRST NAME
Birth name
Address
ZIP codeTown
Date of birth:

EMERGENCY SITUATION:

- ☐ Ongoing pregnancy ☐ Prenatal diagnosis ☐ Pediatric resuscitation

PRESCRIBER

Stamp

Provider identifier (mandatory):

Email address:

Signature:

REQUESTED TEST

IN CASE OF EMERGENCY SITUATION A TRIO ANALYSIS IS NECESSARY
(one form per sample if TRIO analysis)

• **FULL EXOME ANALYSIS (WES)** (SNV/DELINS et CNV) (≈ 22,000 genes + ≈ 12,000 non coding variants (intronic and promoters))

- ☐ SOLO (index case only) (OPL code: EXOME)
☐ SOLO (index case only) (OPL code: EXOME) + segregation study of variant(s) of interest if positive (reflex test) (OPL code: parents ADNGS+10003)
☐ TRIO (index case AND their 2 parents) (OPL code: index case TRIO, parents TRIOP)

• **NGS PANEL ANALYSIS*** (SNV/DELINS and CNV)

- ☐ SOLO (index case only)
☐ SOLO (index case only) (OPL code: EXOME) + segregation study of variant(s) of interest if positive (reflex test) (OPL code: parents ADNGS+10003)
☐ TRIO (index case AND their 2 parents)

- ☐ Intellectual disability consensus (190 genes) OPL code: IS062
☐ Epilepsy consensus (306 genes) OPL code: IS043
☐ Inborn errors of metabolism consensus (312 genes) OPL code: IS072
☐ Hereditary pancreatitis (16 genes) OPL code: IS057
☐ Monogenic diabetes (83 genes) OPL code: IS074
☐ Kidney diseases consensus (305 genes) OPL code: IS093
☐ Female infertility (204 genes) OPL code: IS047
☐ Male infertility (193 genes) OPL code: IS070
☐ Other panel* :
- ☐ Hereditary connective tissue disorders (144 genes) OPL code: IS035
☐ Neurologic diseases consensus (380 genes) OPL code: IS079
☐ Neuromuscular diseases (479 genes) OPL code: IS080
☐ Primary immune deficiencies consensus (435 genes) OPL code: IS061
☐ Vision disorder consensus (304 genes) OPL code: IS044
☐ Hereditary hearing loss (436 genes) OPL code: IS051
☐ Noonan syndrome and RASopathies (55 genes) OPL code: IS082
☐ Constitutional bone diseases (310 genes) OPL code: IS108

*A complete description of the panels (>100) and the specific medical prescription forms associated is available on our online catalog.

<https://www.lab-cerba.com/en/catalog> (key words: NGS PANEL)

You can request a list of genes (polegenetmol@lab-cerba.com)

• **SINGLE GENE ANALYSIS** (OPL code: MGDMD) / **CUSTOM PANEL** (address your request to: polegenetmol@lab-cerba.com)

HGNC if applicable

Indicate the name of the gene to study and its

• **TARGETED VARIANT TEST** (OPL code: MGMUT) (only as part of a family study or for NGS confirmation)

It is **essential** the index case report is attached, otherwise indicate the Cerba file identification number if it was performed at our laboratory:

Indicate the name of the variant to test for

ALREADY PERFORMED TESTS

- ☐ Karyotype / Fish ☐ Array CGH / CMA ☐ Mitochondrial test
☐ Tested gene or genes panel: ☐ Other test(s)

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CLINICAL INFORMATION

Symptomatic patient ☐ Yes ☐ No **If yes: symptoms appeared at years old**
Diagnostic hypothesis:
 Please indicate main clinical signs:

MOST RELEVANT INDICATION

Development of anomalies and intellectual disability Neurodevelopment conditions	Motor or cognitive conditions of the central nervous system
☐ Development anomaly with diagnostic hypothesis ☐ Development anomaly with no diagnostic hypothesis ☐ Intellectual disability with diagnostic hypothesis ☐ Intellectual disability with no diagnostic hypothesis ☐ Autism spectrum disorder ☐ Cerebral deformity (except cerebellum and brainstem deformities) ☐ Cerebellum and brainstem deformities ☐ Epilepsy ☐ Other:	☐ Fronto-temporal dementia ☐ Dystonia ☐ Leukodystrophy and Leucoencephalopathy ☐ Dominant autosomal Alzheimer's disease ☐ Parkinson's disease ☐ Hemiplegic migraine ☐ Abnormal movements ☐ Hereditary spastic paraplegia ☐ Other:
Bone, calcium and cartilage conditions Head, neck and teeth conditions	Neuromuscular conditions
☐ Overgrowth syndrome ☐ Cerebellum and brainstem deformities ☐ Polydactylie - Syndactylie - Triphalangie ☐ Statural delay ☐ Limb deformity with no diagnosis ☐ Calcium-phosphate metabolism anomaly ☐ Isolated cleft lip or palate ☐ Syndromic cleft lip or palate (including Pierre Robin's syndrome) ☐ Other:	☐ Mitochondrial condition via mutation of nuclear genes ☐ Limb-girdle muscular dystrophy ☐ Duchenne and Becker's muscular dystrophy ☐ Unlabeled myopathy ☐ Congenital myopathy ☐ Congenital myasthenic syndrome ☐ Congenital hypotonia ☐ Charcot-Marie-Tooth disease ☐ Sensitive and autonom hereditary peripheral neuropathy ☐ Amyotrophic lateral sclerosis and other rare conditions of the motoneuron ☐ Other:
Hereditary metabolism conditions Rare liver conditions in children and adults	Sensory conditions
☐ Familial hypertriglyceridemia ☐ Familial hypercholesterolemia (hyperLDLemia or hyper- β -lipoproteinemia) ☐ Glycogen storage disease ☐ Peroxisomal pathology ☐ Lysosomal overload condition ☐ Organic aciduria ☐ Aminoacidopathy ☐ Other:	☐ Isolated or syndromic neurosensory hearing loss ☐ Isolated or syndromic hereditary retinal dystrophy ☐ Isolated or syndromic ocular development anomaly ☐ Isolated or syndromic congenital cataract ☐ Isolated or syndromic hereditary corneal dystrophy ☐ Albinism ☐ Hereditary optic neuropathy (HON) ☐ Other:
Immuno-hematologic conditions	Endocrine conditions
☐ Hereditary immunodeficiency with no diagnostic hypothesis ☐ Immunodeficiency impacting humoral immunity (lack of antibody production) ☐ Immunodeficiency impacting cellular and humoral immunity ☐ Neonatal neutropenia ☐ Other:	☐ Non obstructive azoospermia ☐ Obstructive azoospermia unrelated to the absence of deferent ducts ☐ Rare feminine infertility ☐ Isolated premature ovarian failure ☐ Monogenic diabetes ☐ Neonatal diabetes ☐ Hyperinsulinism ☐ Other:
Dermatologic conditions	Vascular conditions with multisystemic impact
☐ Albinism ☐ Ichtyosis ☐ Tuberous sclerosis complex ☐ Incontinentia pigmenti ☐ Hereditary epidermolysis bullosa ☐ Other:	☐ Marfan's syndrome and similar pathologies ☐ Vascular Ehlers-Danlos syndrome ☐ Hereditary hemorrhagic telangiectasia ☐ Other:
Other	
☐ Hereditary and idiopathic pancreatitis ☐ Ciliary dyskinesia ☐ Other :	

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FAMILY INFORMATION

Consanguinity ☐ Yes ☐ No
 Sibling death ☐ Yes ☐ No
 Affected twins ☐ Yes ☐ No

FAMILY TREE

☐ Man
☐ Woman
☐ Unknown sex
☒ ☒ ☒ Affected person
☐ ☐ ☐ Healthy person

PATIENT'S MOTHER

2 tubes of 5 ml whole blood in EDTA

SURNAME
 FIRST NAME
 Birth name
 Address.....
 ZIP code Town
 Date of birth:
 Date of collection:
 Same clinical presentation as index case patient:
☐ Yes ☐ No (include clinical description)

PATIENT'S FATHER

2 tubes of 5 ml whole blood in EDTA

SURNAME
 FIRST NAME
 Address
 ZIP code Town
 Date of birth:
 Date of collection:
 Same clinical presentation as index case patient:
☐ Yes ☐ No (include clinical description)