Cerba

MEDICAL PRESCRIPTION FORM

MOLECULAR DIAGNOSIS OF A GENETIC DISEASE BY NGS

EXOME AND GENE PANELS

Laboratoire Cerba
Customer relation service
Tel.: +33 (0)1 34 40 97 76
Fax: +33 (0)1 34 40 21 29
Email: intgb@lab-cerba.com

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 villositis
- ☐ Chorionic villositis (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 code .....Town .....

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 SITUATON 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 (ndex 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 erors 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 infertiliy (193 genes) *OPL code: IS070*
- ☐ 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*
- ☐ Other panel* :.....

*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: MGDM0)* / **CUSTOM PANEL** (address your request to: polegenetmol@lab-cerba.com)

HGNC if applicable

Indicate the name ot the gene to study and its

● **TARGETED VARIANT TEST** *(OPL code: MGMUT)* (exclusively in the context of a family study or for NGS confirmation)

Attach the report of the Index Case or the Relative mentioning the variant to be searched (HGVS nomenclature) or indicate the Cerba file number if performed by our laboratory.

Gene(s) to be studied: ..... **Reference** sequence: ..... **Variant**(s) to be analyzed: .....

CERBA file number / index case or relative: .....

Note: The index case must be tested for any family segregation request

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

- ☐ 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: .....

Motor or cognitive conditions of the central nervous system

- ☐ 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

- ☐ 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: .....

Neuromuscular conditions

- ☐ 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

- ☐ Familial hypertriglyceridemia
- ☐ Familial hypercholesterolemia (hyperLDLemia or hyper-β-lipoproteinemia)
- ☐ Glycogen storage disease
- ☐ Peroxisomal pathology
- ☐ Lysosomal overload condition
- ☐ Organic aciduria
- ☐ Aminoacidopathy
- ☐ Other: .....

Sensory conditions

- ☐ 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 neurotpathy (HON)
- ☐ Other: .....

Immuno-hematologic conditions

- ☐ Hereditary immunodeficiency with no diagnostic hypothesis
- ☐ Immunodeficiency impacting humoral immunity (lack of antibody production)
- ☐ Immunodeficiency impacting cellular and humoral immunity
- ☐ Neonatal neutropenia
- ☐ Other: .....

Endocrine conditions

- ☐ 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

- ☐ Albinism
- ☐ Ichtyosis
- ☐ Tuberous sclerosis complex
- ☐ Incontinentia pigmenti
- ☐ Hereditary epidermolysis bullosa
- ☐ Other: .....

Vascular conditions with multisystemic impact

- ☐ 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)*