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 EXADP+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 EXADP+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)* **(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

- ☐ 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)*