MEDICAL PRESCRIPTION FORM

CONSULTATION CERTIFICATE / CERTIFICAT OF INFORMATION AND CONSENT FOR TESTING

Laboratoire Cerba
Customer relation service
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GENETIC DISEASE – MOLECULAR DIAGNOSTIC BY NGS
EXOME – GENE PANEL- CUSTOM PANEL
NEUROLOGY

SAMPLING (one form per sample if request for a TRIO analysis)

Sampling date: |_|_|/|_|_|/|_|_|_|_| **Customer**: |_|_|_|_|_| / |_|_|

PRENATAL DIAGNOSIS (*check the corresponding box; a maternal blood sample in a 5-mL EDTA whole blood tube* must be enclosed) for contamination study:

☐ Amniotic fluid (FRESH)	☐ Amniotic fluid (CULTURE)	☐ Fetal DNA
☐ Chorionic villi	☐ Chorionic villi (CULTURE)	☐ Fetal blood
POSTNATAL DIAGNOSIS:	☐ EDTA whole blood	☐ DNA
FETOPATHOLOGY:	☐ Fetal biopsy	☐ Fetal DNA

PATIENT

LAST NAME .....
FIRST NAME .....
Birth name .....
Address .....
City ..... Country .....
Date of birth: |_|_|/|_|_|/|_|_|_|_|
Country of origin: .....

EMERGENCY:

☐ Ongoing pregnancy ☐ Prenatal diagnosis ☐ Pediatric resuscitation

PRESCRIBER

LAST NAME .....
FIRST NAME .....
Address .....
City ..... Country .....
Tel.: .....
Fax: .....
Email address: .....

Signature:

REQUESTED TEST - IN CASE OF PRENATAL DIAGNOSIS OR NEONATAL RESUSCITATION: A TRIO ANALYSIS IS REQUIRED
(One form per sample if request for a TRIO analysis)

● **EXOME ANALYSIS** (Includes the analysis of SNV/insdel and CNV)

☐ SOLO (Exome analysis only in the index case) *(OPL code: EXOME)*
☐ TRIO (Joint Exome analysis in the index case AND its parents) *(OPL Code: index case TRIO, parents TRIOP)*
☐ SOLO+Segregation (Exome analysis only in the index case +/- Segregation analysis of candidate variant) *(OPL code: index case EXOME, parents ADNGS+10003)*

● **NGS PANEL*** (SNV/insdel and CNV) * *See our online catalogue for the respective sub-panels. Gene list on request* (equipe.mgdm@lab-cerba.com)

☐ Organic Acidemias (46 genes) *OPL code: IS084*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Transthyretin cardiac amyloidosis (1 gène) *OPL code: IS023*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Ataxia Telangiectasia (4 genes) *OPL code: MGDM0*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Charcot-Marie-Tooth (307 genes) *OPL code: IS026*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Intellectual Disability MEDIANE CONSENSUS (190 genes) *OPL code: IS062*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Dyskeratosis Congenita (20 genes) *OPL code: IS039*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Septo-optical Dysplasia (19 genes) *OPL code: IS096*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Epilepsy MEDIANE CONSENSUS (306 genes) *OPL code: IS043*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Leukodystrophy (389 genes) *OPL code: IS066*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Lysosomal Disorders (80 genes) *OPL code: IS068*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Parkinson (168 genes) *OPL code: IS086*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Migraine (29 genes) *OPL code: IS073*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Spastic Paraplegia (172 genes) *OPL code: IS100*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Amyotrophic Lateral Sclerosis (76 genes) *OPL code: IS004*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Joubert and Meckel Syndrome (51 genes) *OPL code: IS063*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Noonan and RASopathies (55 genes) *OPL code: IS082*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Hereditary Hemorrhagic Telangiectasia (6 genes) *OPL code: IS055*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation
☐ Neurology MEDIANE CONSENSUS Comprehensive Panel (380 genes) *OPL code: IS079*	☐ SOLO	☐ TRIO	☐ SOLO+Segregation

☐ **SINGLE GENE ANALYSIS** *(OPL code: MGDM0)* / **CUSTOM PANEL** (send your request to: equipe.mgdm@lab-cerba.com)

Enter the name of the gene to be studied and its HGNC symbol

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

Enter the name of the variant to be detected and enclose the index case report

TESTS ALREADY PERFORMED PRIOR TO THIS TEST

☐ Karyotype / Fish	☐ CGH-Array / ACPA	☐ Mitochondrial test
☐ Gene or gene panel tested:		☐ Other test(s) .....

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EXOME – GENE PANEL- CUSTOM PANEL

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INDICATION

Symptomatic patient ☐ Yes ☐ No If yes, age at symptom onset: ..... years

Clinical suspicion: .....

Symptoms (check all the information in the table below):

PERINATALITY

- ☐ Preterm birth (HPO: HP:0001622)
- ☐ Intrauterine growth retardation (HPO: HP:0001511)
- ☐ Oligohydramnios (HPO: HP:0001562)
- ☐ Polyhydramnios (HPO: HP:0001562)
- ☐ Cystic hygroma (HPO: HP:0000476)
- ☐ History of hydrops fetalis (HPO: HP:0012050)
- ☐ Other: .....

GROWTH

- ☐ Failure to thrive (HPO: HP:0004322)?
- ☐ Overgrowth (HPO: HP:0000098)?
- ☐ Other: .....

COGNITIVE

- ☐ Developmental delay (HPO: HP:0001263)
- ☐ Fine motor disorder (HPO: HP:0010862)
- ☐ General motor disorder (HPO: HP:0002194)
- ☐ Speech disorder (HPO: HP:0000750)
- ☐ Cognitive impairment (HPO: HP:0001249)
- ☐ IQ: .....
- ☐ Developmental regression (HPO: HP:0002376)
- ☐ Other: .....

BEHAVIOR

- ☐ Autism (HPO: HP:0000717)
- ☐ Pervasive developmental disorder (PDD) (HPO: HP:0000708)
- ☐ Hyperactivity (HPO: HP:0000752)
- ☐ Anxiety (HPO: HP:0000739)
- ☐ Self-injury (HPO: HP:0000742)
- ☐ Other: .....

MUSCULOSKELETAL

- ☐ Clubfoot (HPO: HP:0001762)
- ☐ Diaphragmatic hernia (HPO: HP:0000776)
- ☐ Polydactyly (HPO: HP:0010442)
- ☐ Clinodactyly (HPO: HP:0030084)
- ☐ Syndactyly (HPO: HP:0001159)
- ☐ Clenched hands (HPO: HP:0001188)
- ☐ Talus verticalis (HPO: HP:0001838)
- ☐ Contractures (HPO: HP:0001371)
- ☐ Scoliosis (HPO: HP:0002650)
- ☐ Joint stiffness/limitation (HPO: HP:0002063)
- ☐ Marfanoid appearance (HPO: HP:0001519)
- ☐ Osteopenia (HPO: HP:0000938)
- ☐ Osteoporosis (HPO: HP:0000939)
- ☐ Other: .....

INFERTILITY

- ☐ Non-obstructive azoospermia (HPO: HP:0011961)
- ☐ Teratozoospermia (HPO: HP:0012864)
- ☐ Premature ovarian failure* (HPO: HP:008209)
- ☐ Other: .....

*According to the ESHRE criteria: onset before the age of 40, amenorrhea for more than 4 months associated with a FSH level >25 mIU/mL on at least two samples and decreased estradiol level

CRANIOFACIAL / OPHTHALMOLOGY / HEARING

- ☐ Macrocephaly (HPO: HP:0000256)
- ☐ Microcephaly (HPO: HP:0000252)
- ☐ Cleft lip and palate (HPO: HP:0000175)
- ☐ Macroglossia (HPO: HP:0000158)
- ☐ Craniosynostosis (HPO: HP:0001363)
- ☐ Abnormality of the philtrum (HPO: HP:0000288)
- ☐ Facial hypoplasia (HPO: HP:0000274)
- ☐ Irregular teeth (HPO: HP:0040079)
- ☐ Cataract (HPO: HP:0000518)
- ☐ Corneal opacity (HPO: HP:0007957)
- ☐ Lens dislocation (HPO: HP:0001083)
- ☐ Cherry-red spot in the macula (HPO: HP:0010729)
- ☐ Retinitis pigmentosa (HP:0000510)
- ☐ Nystagmus (HPO: HP:0000639)
- ☐ Ophthalmoplegia (HPO: HP:0000602)
- ☐ Coloboma (HPO: HP:0000589)
- ☐ Ptosis (HPO: HP:0000508)
- ☐ Strabismus (HPO: HP:0000486)
- ☐ Blindness (HPO: HP:0000618)
- ☐ Preauricular appendage (HPO: HP:0000384)
- ☐ Microtia (HPO: HP:0008551)
- ☐ Outer ear deformity (HPO: HP:0000356)
- ☐ Hearing loss or deafness (HPO: HP:0000365)
- ☐ Facial dysmorphia (HPO: HP:0001999)

  Description: .....

  .....
- ☐ Other: .....

CARDIAC

- ☐ AVSD (HPO: HP:0006705)
- ☐ VSD (HPO: HP:0010438)
- ☐ Aortic coarctation (HPO: HP:0001680)
- ☐ Hypoplastic left heart syndrome (HPO: HP:0004383)
- ☐ Tetralogy of Fallot (HPO: HP:0001636)
- ☐ Transposition of the great vessels (HPO: HP:0001669)
- ☐ Cardiomyopathy (HPO: HP:0001638)
- ☐ Other: .....

NEUROMUSCULAR

- ☐ Ataxia (HPO: HP:0001251)
- ☐ Chorea (HPO: HP:0002072)
- ☐ Exercise intolerance (HPO: HP:0003546)
- ☐ Fatigue (HPO: HP:0012378)
- ☐ Headaches/migraines (HPO: HP:0002076)
- ☐ Dystonia (HPO: HP:0001332)
- ☐ Hypotonia (HPO: HP:0001290)
- ☐ Hypertonia (HPO: HP:0001276)
- ☐ Spasticity (HPO: HP:0001257)
- ☐ Paraplegia (HPO: HP:0010550)
- ☐ Reye syndrome/Pseudo-Reye syndrome (HP:0006582)
- ☐ History of stroke (HPO: HP:0002401)
- ☐ Neuropathy (HPO: HP:0009830)
- ☐ Epilepsy/Seizure (HPO: HP:0001250)
- ☐ Other: .....

IMMUNITY

- ☐ Recurrent infections (HPO: HP:0002719)
  - ○ Types of infections: .....
  - ○ Frequency/year: .....
  - ○ Pathogens involved: .....
- ☐ Other manifestations: .....

METABOLIC

- ☐ Lucid interval
- ☐ Ketosis (HPO: HP:0001946)
- ☐ Lactic acidosis (HPO: HP:0003128)
- ☐ Hyperammonemia (HPO: HP:0001987)
- ☐ Hyperuricemia (HPO: HP:0002149)
- ☐ Hypoglycemia (HPO: HP:0001943)
- ☐ Hyperglycemia (HPO: HP:0003074)
- ☐ Organic aciduria (HPO: HP:0001992)
- ☐ Other: .....

HEMATOLOGY/IMMUNOLOGY

- ☐ Anemia (HPO: HP:0001903)
- ☐ Neutropenia (HPO: HP:0001875)
- ☐ Pancytopenia (HPO: HP:0001876)
- ☐ Blood clotting disorder (HPO: HP:0001928)
- ☐ Autoimmune disease (HPO: HP:0002960)
- ☐ Other: .....

GASTROINTESTINAL

- ☐ Jaundice (HPO: HP:0000952)
- ☐ Vomiting (HPO: HP:0002013)
- ☐ Feeding difficulties (HPO: HP:0011968)
- ☐ Gastroschisis (HPO: HP:0001543)
- ☐ Omphalocele (HPO: HP:0001539)
- ☐ Anal atresia (HPO: HP:0002023)
- ☐ Tracheoesophageal fistula (HPO: HP:0002575)
- ☐ Hepatomegaly (HPO: HP:0002240)
- ☐ Splenomegaly (HPO: HP:0001744)
- ☐ Hepatocellular failure (HPO: HP:0001399)
- ☐ Hyperechogenic fetal colon
- ☐ Pyloric stenosis (HPO: HP:0002021)
- ☐ Other: .....

ENDOCRINOLOGY

- ☐ ☐ Type I ☐ Type II diabetes
- ☐ Hypothyroidism (HPO: HP:0000821)
- ☐ Hypoparathyroidism (HPO: HP:0000829)
- ☐ Hyperparathyroidism (HPO: HP:0000843)
- ☐ Other: .....

UROGENITAL TRACT

- ☐ Sexual ambiguity (HPO: HP:0000062)
- ☐ Hypospadia (HPO: HP:0000047)
- ☐ Cryptorchidism (HPO: HP:0000028)
- ☐ Kidney malformation (HPO: HP:0000077)
- ☐ Renal agenesis (HPO: HP:0000104)
- ☐ Hydronephrosis (HPO: HP:0000126)
- ☐ Renal cysts (HPO: HP:0000107)
- ☐ Tubulopathy (HPO: HP:0000114)
- ☐ Nephropathy (HPO: HP:0000112)
- ☐ Hypohidrosis (HPO: HP:0000966)
- ☐ History of lithiasis: if yes, nature? .....
- ☐ Other: .....

BRAIN ABNORMALITY

- ☐ Dandy-Walker malformation (HPO: HP:0001305)
- ☐ Holoprosencephaly (HPO: HP:0001360)
- ☐ Lissencephaly (HPO: HP:0001339)
- ☐ Agenesis of the corpus callosum (HPO: HP:0001274)
- ☐ Hydrocephalus (HPO: HP:0000238)
- ☐ Involvement of the basal ganglia (HPO: HP:0002134)
- ☐ Hypomyelination (HPO: HP:0003429)
- ☐ Demyelination (HPO: HP:0007305)
- ☐ Cerebellar atrophy (HPO: HP:0007360)
- ☐ Ventricular dilation (HPO: HP:0002119)
- ☐ Other: .....

Other relevant clinical information: .....

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FAMILY INFORMATION

Consanguinity ☐ Yes ☐ No

Death in siblings ☐ Yes ☐ No

Affected twins ☐ Yes ☐ No

FAMILY TREE

☐ Man
○ Woman
◇ Individual of unknown sex
■ ● ◆ Affected subject
☐ ○ ◇ Healthy subject

MOTHER OF THE PATIENT 2 x 5-mL EDTA tubes of whole blood

LAST NAME .....

FIRST NAME .....

Birth name .....

Address .....

City ..... Country .....

Date of birth: |__|__|__|__|__|__|__|__|

Sampling date: |__|__|__|__|__|__|__|__|

Sample taken to:

☐ only check identified variants in the index case

☐ an exome analysis (note: invoicing for a trio analysis in this case)

Same clinical presentation as the index case patient:

☐ Yes ☐ No *(enclose the clinical description)*

FATHER OF THE PATIENT 2 x 5-mL EDTA tubes of whole blood

LAST NAME .....

FIRST NAME .....

Address .....

City ..... Country .....

Date of birth: |__|__|__|__|__|__|__|__|

Sampling date: |__|__|__|__|__|__|__|__|

Sample taken to:

☐ only check identified variants in the index case

☐ an exome analysis (note: invoicing for a trio analysis in this case)

Same clinical presentation as the index case patient:

☐ Yes ☐ No *(enclose the clinical description)*

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CONSULTATION CERTIFICAT FROM THE PRESCRIBER
CERTIFICAT OF INFORMATION AND CONSENT PATIENT FOR TESTING

GENETIC DISEASE – MOLECULAR DIAGNOSTIC BY NGS
EXOME – GENE PANEL – CUSTOM PANEL

The signed consultation certificate and consent must be enclosed (Document below)

CONSULTATION CERTIFICATE FROM THE PRESCRIBING PHYSICIAN OR THE GENETIC COUNSELOR

☐ **POSTNATAL DIAGNOSIS**

I, the undersigned, Dr./Prof. …… or …… genetic counselor under the supervision of Dr./Prof. …… certify that I have informed the undersigned patient and his/her parents (legal representatives) about the characteristics of the investigated disease, how to diagnose it, how to prevent and treat it, how the disease is transmitted and the possible consequences in other members of the family, the storage of the sample, and that I have obtained the consent of the patient AND his/her guardianship under the conditions provided for by the French public health code (articles R113-4 and 5).

☐ **PRENATAL DIAGNOSIS**

I, the undersigned, Dr./Prof. …… or …… genetic counselor under the supervision of Dr./Prof. …… certify that I have informed the undersigned patient about the risk to her child of being affected by a particularly serious chromosomal, genetic or infectious abnormality, the characteristics of this disease, how to detect it, the associated risk and the possible consequences of an abnormal outcome.

CONSENT OF THE PREGNANT WOMAN FOR SAMPLE COLLECTION AND TESTING FOR AN *IN UTERO* PRENATAL DIAGNOSIS

Decree of January 14, 2014, consolidated on January 2, 2019

I, the undersigned, Mrs. …… certify that I have received:

- Information on the risk to the unborn child of being affected by a particularly serious disease, the characteristics of this disease; how to diagnose it; the potential options of fetal medicine, treatment or management of the born child.
- Information on laboratory tests likely to allow making an in utero prenatal diagnosis that have been offered to me and that I would like to perform: this or these tests require the collection of a sample of amniotic fluid, chorionic villi (placenta), fetal blood or any other fetal sample; the procedures, risks, disadvantages and possible consequences of each sampling technique necessary to perform this or these tests have been explained to me; I have been informed that a second sample may be required in case of technical failure; if this happens, I will have to sign a new written consent; other diseases than that or those initially investigated could be revealed by the test; I have been informed that the result of the test will be available to me and explained to me by the physician who prescribed it.

I consent to the collection (required for testing) of (*):

☐ amniotic fluid ☐ chorionic villi

☐ fetal blood ☐ other fetal sample (specify)……

I also consent to the test(s) (*) for which this sample is taken:

☐ cytogenetic testing, including molecular tests applied to cytogenetics;

☐ molecular genetic testing;

☐ fetal chemistry diagnostic tests;

☐ laboratory tests for the diagnosis of infectious diseases.

CONSENT FOR GENETIC TESTING OF A PERSON

- Information on the genetic test that is offered to me, that will be performed on (check below):
  - ☐ the biosample(s) taken from me
  - ☐ the biosample(s) taken from my child or from the adult under guardianship
  - ☐ the sample that will be taken from my dead fetus
- Information on the genetic tests that will be performed to:
  - confirm or rule out the diagnosis of a genetic disease related to my symptoms;
  - confirm or rule out the presymptomatic diagnosis of a genetic disease;
  - identify a healthy carrier status (screening for heterozygous variants or chromosomal rearrangement)
  - assess my genetic susceptibility to a disease or drug treatment.

I have been informed:

- Of my right to request the interruption of this study, that the results are not communicated to me, or the destruction of the stored samples
- That the full interpretation of these results is based, in some situations, on the definition of biological relationships, which can be analyzed from these samples.
- Of my responsibility regarding my duty to inform my family, if a serious genetic abnormality is revealed, the consequences of which are likely to result in the implementation preventive measures, including genetic counseling or care.

I authorize the storage of a biosample taken to me and its subsequent use to **continue investigations as part of the same diagnostic process**, depending on the evolution of knowledge.	☐ Yes ☐ No
The technique used may **reveal genetic information that is unrelated to the investigated disease, but that may have an impact on my health or that of relatives**. I would like to be informed of these results.	☐ Yes ☐ No
I authorize the transmission of a sample along with the necessary medical data, including any photographs, to another laboratory to **complete this genetic study**. I authorize the **recording and storage of medical data** useful for the management of the diagnostic process in computer databases	☐ Yes ☐ No
I authorize the **recording and storage of medical data** useful for the management of the diagnostic process in computer databases	☐ Yes ☐ No
As part of the diagnostic process, part of my sample may not be used. I authorize its storage and use for internal laboratory quality assurance studies.	☐ Yes ☐ No
I authorize the anonymized use of medical data and/or part of the samples within the framework of research projects, of a scientific study program without direct benefit or loss to me (all my medical data will be protected through total anonymization).	☐ Yes ☐ No

The result of this test will be available to me and explained to me by the prescribing physician (or by the delegated genetic counselor) in the current state of knowledge in the context of a genetic consultation. This or these tests will be performed by a medical biology laboratory authorized by the regional health agency to perform them. The original of this document will be kept in my medical record. A copy of this document will be provided to me and to the practitioner who must perform the tests. The medical biology laboratory in which the practitioner who performed the tests works will keep this document under the same conditions as the test report. I have had the opportunity to ask questions to the geneticist or genetic counselor who prescribed this test and all my questions have been answered satisfactorily.

Done in …… on ……

PATIENT ID (*Signature*)	LEGAL REPRESENTATIVE ID (*Signature*)	PRESCRIBER (*Signature*)
Last name: *First name:* *Date of Birth:*	Father (*first and last name, date of birth*): Mother (*first and last name, date of birth*): If the patient is minor or an adult under guardianship, relationship to the patient:	*Last name:* *First name:*

Cerba Laboratory is responsible for processing your personal data provided in this form as the data controller to perform tests, interpret them, transmit results, and manage the laboratory's administrative tasks. If you are a patient, your data may then be reused for anonymization purposes for scientific research, quality control, statistical studies, or satisfaction surveys. To learn more about how your personal data is processed, your rights, and research projects based on your data, visit the website www.lab-cerba.com, under the "Personal Data" and "Research Projects" tabs. To exercise your rights of opposition, contact our Data Protection Officer (DPO) at: rpd.cerba@labcerba.com / CERBA–RPD – ZAC DES EPINEAUX 10-12 Avenue ROLAND MORENO CS 51312 95740 FREPILLON. / If you are a patient, in accordance with applicable regulations, once your tests are completed, any remaining samples from your tests will be disposed of. However, they may be kept for use in scientific research or quality control purposes, either directly or after transfer to third parties, in strict compliance with medical confidentiality. You may object to such use by simply making a request to our DPO (contact details above).