MEDICAL PRESCRIPTION FORM

CONSULTATION CERTIFICATE / CONSENT TESTING

DEVELOPMENTAL DISORDERS AND GENETIC DISEASES

Conventional Cytogenetics, Molecular Cytogenetics, and Molecular Genetics

Please ensure the completed and signed consultation certificate and consent form are attached (document on page 3)

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

IMPORTANT

- For the following tests (not included in this document), please refer to the dedicated prescription forms available on MyCerba or the website https://www.lab-cerba.com:
- Reproductive Disorder (Karyotype, Gene Study, or Gene Panel Analysis)
- Cystic Fibrosis and Associated Pathologies (CFTR Gene or Gene Panel Analysis)
- SHOX Gene Study
- Noonan Syndrome and RASopathies (PTPN11 Gene or Gene Panel Analysis)
- Rett Syndrome (*MECP2 gene*)
- Hereditary Auto-Inflammatory Disease (Gene Study or Gene Panel Analysis): Familial Mediterranean Fever ; TRAPS Syndrome ; Hereditary Periodic Fever Syndrome linked to NLRP12 ; Periodic Fever Syndrome with Hyper-IgD
- Globin Genes (Sickle Cell Disease, Beta-Thalassemia, Variant Studies)

LABORATORY AND SAMPLE COLLECTION

Client number ⌊_⌋⌊_⌋⌊_⌋⌊_⌋⌊_⌋ / ⌊_⌋ **Date of sample collection** ⌊_⌋⌊_⌋/⌊_⌋⌊_⌋/⌊_⌋⌊_⌋⌊_⌋⌊_⌋ **Sample time** ⌊_⌋⌊_⌋ **h** ⌊_⌋⌊_⌋

Sample Type ☐ Total Blood EDTA ☐ Total Blood Heparinized **Number of tubes**: ⌊_⌋

FOETOPATHOLOGY : ☐ Tissue (in culture medium), please specify.....

PATIENT

NAME .....

FIRST NAME .....

Birth name.....

Address .....

Zip Code ..... Town .....

Date of birth : ⌊_⌋⌊_⌋/⌊_⌋⌊_⌋/⌊_⌋⌊_⌋⌊_⌋⌊_⌋

EMERGENCY SITUATION:

☐ Ongoing pregnancy ☐ Prenatal Diagnostic ☐ Pediatric Resuscitation

PRESCRIBER

Dr .....

Mandatory Stamp

Email address : .....

Signature :

INFORMATION ON RELATIVE

SPOUSE :	LAST NAME	FIRST NAME	Date of birth	⌊_⌋⌊_⌋/⌊_⌋⌊_⌋/⌊_⌋⌊_⌋⌊_⌋⌊_⌋
FATHER :	LAST NAME	FIRST NAME	Date of birth	⌊_⌋⌊_⌋/⌊_⌋⌊_⌋/⌊_⌋⌊_⌋⌊_⌋⌊_⌋
MOTHER :	LAST NAME	FIRST NAME	Date of birth	⌊_⌋⌊_⌋/⌊_⌋⌊_⌋/⌊_⌋⌊_⌋⌊_⌋⌊_⌋

CYTOGENETICS AND MOLECULAR CYTOGENETICS : TESTS AND INDICATIONS

REQUESTED TESTS

☐ Constitutional Karyotype on blood from patients over 8 days old (heparinized tube) *(OPL: 09703)*

☐ Constitutional Karyotype on blood from newborns (0 to 8 days old) (heparinized tube) *(OPL: 09709)*

☐ Chromosomal Analysis by DNA Microarray (ACPA) (EDTA tube) *(OPL: PPOST)*

☐ Chromosomal Analysis by DNA Microarray (ACPA) for Family Investigation *(EDTA tube) (OPL : PPARE)* : Cerba case number of the index case: .....

☐ Search for a Microdeletion Syndrome (FISH technique) (heparinized tube) :

☐ Wolf-Hirschhorn (4p-) ☐ Cri du Chat (5p-) ☐ Willi-Prader ☐ Angelman ☐ Williams-Beuren

☐ Smith-Magenis ☐ Miller-Diecker ☐ DiGeorge ☐ Other: .....

For an Optical Genome Mapping (OGM): Please complete the dedicated prescription form available for download on **MyCerba or on the website** https://www.lab-cerba.com

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INDICATION (cytogentic and molecular cytogenetic examination)

- ☐ Intellectual disability or learning disorders in a syndromic context (indc62). Specify ……
- ☐ Suspected Down syndrome (indc22)
- ☐ Malformations WITHOUT psychomotor delay (indc63) Specify ……
- ☐ Isolated intellectual disability or learning disorders (indc64) Specify ……
- ☐ Pervasive developmental disorders (PDD)/autism, or psychiatric/behavioral disorders (indc65)
- ☐ Isolated Epilepsy (indc40)
- ☐ Growth disorders, specify:
  - ☐ Suspected Turner syndrome (indc27) ☐ Height advancement (indc59) ☐ Height delay (indc18)
- ☐ Puberty advancement (indc61) ☐ Puberty delay (indc19)
- ☐ Primary amenorrhea (indc5) ☐ Secondary amenorrhea (indc6)
- ☐ Suspected Klinefelter syndrome (indc2)
- ☐ Variations in genital development (including secondary) (indc17) Specify ……
- ☐ Family study (indc29) ***(attach results of the index case or contact details of the laboratory that performed the karyotype),*** Specify :
  - ☐ Ongoing prenatal diagnosis ☐ Family study
- ☐ Gamete and embryo donation (indc72)
- ☐ Search for a constitutional anomaly following a somatic examination (indc73) Specify ……
- ☐ Fetopathology examination (indc74)
- ☐ Other (indc23) Specify :……

MOLECULAR GENETICS: TESTS AND INDICATIONS

(Outside of Exome and Gene Panels which have dedicated prescription forms)

REQUESTED TESTS

- ☐ Achondroplasia*[1] (G380R variant if the *FGFR3 gene*)
- ☐ Hypochondroplasia*[1] (N540K and N540S of the *FGFR3 gene*)
- ☐ Thanatophoric dysplasia*[1] (G380R variant of the *FGFR3 gene*)
- ☐ Apert syndrome (G380R variant of the *FGFR2 gene*)

- ☐ Mitochondrial hearing loss
- ☐ Mitochondrial diabetes and hearing loss
- ☐ Leber's hereditary optic neuropathy
- ☐ Mitochondrial cytopathy MERRF
- ☐ Mitochondrial cytopathy MELAS
- ☐ Mitochondrial cytopathy NARP

- ☐ Steinert's myotonic dystrophy
  DMPK gene study (CTG repeat in 3'UTR)
- ☐ Spinal muscular atrophy
  Study of the SMN1/SMN2 gene *(deletion)*

- ☐ Fragile X syndrome
  FMR1 gene study (CGG repeat in 5'UTR)
- ☐ Prader-Willi syndrome (*SNRPN methylation*)
- ☐ Angelman syndrome (*SNRPN methylation*)

- ☐ Search for the SRY gene

- ☐ Tay-Sachs disease: *HEXA* gene study*[2]
- ☐ Canavan disease: *ASPA* gene study*[2]
- ☐ Familial dysautonomia: *IKBKAP* gene study[2]
- ☐ Alpha-1 antitrypsin deficiency: *SERPINA1* gene study
- ☐ Fabry disease: *GLA* gene study[2]
- ☐ Gilbert's disease: *UGT1A1* gene study[2]
- ☐ APOE gene study*[2] :
  - ☐ Familial dyslipoproteinemia
  - ☐ Neurodegenerative disease
- ☐ Nash - PNPLA3 gene: polymorphism c.444G>C
- ☐ Glucose transporter deficiency syndrome:
  SCL2A1 gene study
- ☐ Primary lactose intolerance : LCT gene study

** 1 – Également présent dans le panel NGS Syndrome de petite taille https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/FR/FDE_EXOME_PANELS_ENDOCRINO.pdf*
** 2 – Également présent dans le panel NGS Métabolisme https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/FR/FDE_EXOME_FR_PANELS_METABO.pdf*

INDICATION

- ☐ Index Case Study Please specify clinical suspicion: ……
- ☐ Family Study – Related Case Study Specify: ……
  - ☐ Symptomatic Relative ☐ Asymptomatic Relative ☐ Parent of fetus with ultrsound abnormalities
  - ☐ Parent of fetus for Prenatal Diagnosis (known variant)
- ☐ Heterozygosity Screening Specify: ……
  - ☐ Personal Family History ☐ Spouse's Family History ☐ No Family History ☐ Other: ……
- ☐ Gamete Donation
- ☐ Uniparental Disomy *(OPL : DUP)* Specify: ……

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MEDICAL CONSULTATION CERTIFICATE
PATIENT INFORMATION AND CONSENT CERTIFICATE

GENETIC CHARACTERISTICS EXAMINATION OF AN INDIVIDUAL

Please attach the completed and signed medical consultation certificate and patient information and consent

Physician Prescriber or Genetic Counselor Consultation and Information Certificate

(Section for the Prescriber)

I, the udersigned, Dr./Prof. [*First Name, Last Name*],

Or Ms./Mr. [*First Name, Last Name*] ..... , genetic counselor*

Certify that I received in consultation today :

Ms./Mr. [*First Name, Last Name*]……, Born on [*Date of Birth*]……

Certify that I provided (or provided to the legal guardians or the guardian) all the information mentioned in articles R. 1131-4 and R. 1131-20-1 and following of the Public Health Code as well as in the texts taken for their application :

1. The characteristics of the disease being sought, the means of detecting it, the reliability of the tests, and the possibilities of prevention measures, including genetic counseling, and care;
2. The genetic transmission modalities of the disease being sought when known and their possible consequences for other members of their family ;
3. That the examination may incidentally reveal genetic characteristics unrelated to its initial indication but whose knowledge would allow the person or family members to benefit from prevention measures, including genetic counseling, or care ;
4. That they are required to inform, by any means, potentially affected family members if the diagnosis of this anomaly is confirmed

** In accordance with the provisions of Articles R. 1132-5 and following of the Public Health Code.*

Information and Consent Form for Genetic Testing

(Insert for the patient)

I, undersigned, Mrs./Mr. [*First Name, Last Name*] , attest to having received from the above-mentioned physician during today's medical consultation:

- Information regarding the genetic testing proposed to me, to which I consent, and which will be performed using the biological sample(s) taken [*Check the corresponding box*] :
  - On myself
  - On my minor child or on the adult placed under my guardianship
  - On my stillborn fetus
- Information on the genetic testing to which I consent, and which will be conducted for [*Check the corresponding box*]:
  - Either to establish, confirm, or refute the diagnosis of a genetic disease in a person;
  - Or to identify the characteristics of one or more genes that may lead to the development of a disease in a person or potentially affected family members;
  - Or to tailor medical management of a person based on their genetic characteristics.

I have been informed:

- Of all the points stated in the medical consultation form
- Of my right to withdraw this request for testing(s) at any time, to not receive the results, or to have the stored samples destroyed.
- That the interpretation of these results may, in some situations, rely on the definition of biological parentage, which can be analyzed from these samples.
- Of the procedures for informing family members and my responsibility regarding my duty to inform my family and, if applicable, to communicate with the assisted reproductive technology center in case of gamete donation, if a serious genetic anomaly requiring preventive measures including genetic counseling or care is identified.
- That the result is confidential. It will be provided and explained to me during a consultation by the prescribing physician

I consent to the sampling and the performance of the examination within the scope of [*describe the clinical context*]:

……

……

The technique used may potentially reveal genetic information unrelated to the condition under consideration, but hich may impact my/his/her health or that of relatives, my/his/her management, and/or my/his/her treatment. I wish to be informed of these results..	☐ YES ☐ NO
I agree that if my/his/her results are medically essential for my/his/her relatives, they may be, in accordance ith medical confidentiality, communicated and used anonymously, in their interest, even after my/his/her death.	☐ YES ☐ NO

This (or these) test(s) will be performed in a medical biology laboratory authorized by the Regional Health Agency to conduct them. The original of this document is kept in my medical record. A copy of this document is given to me as well as to the practitioner who will perform the tests. The medical biology laboratory where the practitioner who performed the tests works keeps this document under the same conditions as the examination report. I had the opportunity to ask any questions I wanted to the geneticist or genetic counselor who prescribed this test, and I received complete and adequate answers.

Done at ..... , On .....

PATIENT'S IDENTITY (*Signature*)	IDENTITY of LEGAL REPRESENTATIVE(S)	PRESCRIBER (*Signature*)
Name, first name, date of birth	Signature of both parents required if TRIO Analysis (index case + 2 parents) *Name, first name, date of birth:* *Name, first name, date of birth:* If the patient is a minor or an adult under guardianship, relationship to the patient:	*Name, first name*