Cerba

MEDICAL PRESCRIPTION FORM

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

MOLECULAR DIAGNOSIS OF A NEUROLOGICAL AND NEURODEVELOPMENTAL DISORDER

SAMPLING

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

Customer: |__|__|__|__|__| / |__|__|

POSTNATAL DIAGNOSIS: ☐ EDTA whole blood (from 0.5 ml to 5 ml)

PATIENT

LAST NAME .....

FIRST NAME .....

Maiden Name .....

Address .....

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

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

Mobile phone: .....

E-mail address: .....

PRESCRIBER

Mandatory Stamp

E-mail address: .....

Signature :

REQUESTED TEST

IN CASE OF AN EMERGENCY SITUATION, A TRIO ANALYSIS IS REQUIRED
(One form per sample if request for a TRIO analysis)

● **COMPLETE EXOME ANALYSIS (WES)** (SNV/DELINS and CNV) (≈ 22,000 genes + ≈ 12,000 non-coding variants (intronic and promoter regions)

☐ SOLO (index case only) *(OPL code: EXOME)* ☐ TRIO (index case AND its 2 parents) *(OPL code: index case TRIO,, parents TRIOP)*

☐ SOLO (index case only) *(OPL code: EXOME)* + segregation study of the variant(s) of interest if the result is positive (reflex test) *(OPL code: parents ADNGS+10003)*

● **ANALYSIS OF AN NGS PANEL** (SNV/DELINS and CNV) **OR NUCLEOTIDE EXPANSION**

Neurodegenerative Diseases List of genes available on request (polegenetmol@lab-cerba.com)

- ☐ Amyotrophic Lateral Sclerosis (65 genes) *OPL code: IS004*
- ☐ C9orf72 Amyotrophic Lateral Sclerosis (expansion de l'hexa nucléotide) *OPL code: SLAC9*
- ☐ Dementia and Alzheimer (82 genes) *OPL code: IS133*
- ☐ APOE Genotyping *OPL code: APOEG*
- ☐ Neurodegenerative diseases comprehensive panel (602 genes) *OPL code: IS135*
- ☐ Hereditary Spastic Paraplegia" (217 genes) *OPL code: IS100*
- ☐ Parkinson's Disease (140 genes) *OPL code: IS086*
- ☐ Huntington's Disease (expansion CAG gene *HTT*) *OPL code: HTT*
- ☐ Neurodegeneration with Brain Iron Accumulation (48 genes) *OPL code: IS134*

Neurodevelopmental Disorders List of genes available on request (polegenetmol@lab-cerba.com)

- ☐ Intellectual Disability (2169 genes) *OPL code: IS062*
- ☐ Fragile X Syndrome and FXTAS (FMR1 triplet expansion) *OPL code: FRAXA*
- ☐ Rett Syndrome (*MECP2 gene*) *OPL code: MECP2*
- ☐ Autism Spectrum Disorders (313 genes) *OPL code: IS136*
- ☐ GLUT1 Deficiency Syndrome. MetaGLUT1™ Test *OPL code: GLUDS (see specific request form)*
- ☐ Noonan Syndrome and Rasopathies (55 genes) *OPL code: IS082*
- ☐ Prader-Willi Syndrome (SNRPN methylation) *OPL code: PWSME*
- ☐ Angelman Syndrome (SNRPN methylation) *OPL code: ASSME*
- ☐ Epilepsies (1580 genes) *OPL code: IS043*

Peripheral Neuropathies List of genes available on request (polegenetmol@lab-cerba.com)

- ☐ Familial Amyloid Polyneuropathy (TTR gene) *OPL code: IS137*
- ☐ Hereditary Neuropathies (259 genes) *OPL code: IS138*
- ☐ Charcot-Marie-Tooth Disease (376 genes) *OPL code: IS026*

Central Nervous System Disorders List of genes available on request (polegenetmol@lab-cerba.com)

- ☐ Hereditary Ataxia (409 genes) *OPL code: IS0139*
- ☐ Friedreich Ataxia (FXN triplet expansion) *OPL code: FRTRI*
- ☐ Friedreich Ataxia (coding region sequencing) *OPL code: FRSEQ*
- ☐ Leukodystrophies and Leukoencephalopathies (486 genes) *OPL code: IS066*
- ☐ Dystonia and Abnormal Movements (263 genes) *OPL code: IS140*
- ☐ Hemiplegic Migraine (32 genes) *OPL code: IS141*
- ☐ Brain Malformations (261 genes) *OPL code: IS142*
- ☐ Agenesis of the Corpus Callosum (340 genes) *OPL code: IS143*
- ☐ Central nervous system Vascular Disease (48 genes) *OPL code: IS144*
- ☐ Spinocerebellar Ataxia Panel SCA1,2,3,6,7,12,17 and DRPLA ***OPL code: PSCA***
- ☐ SCA1 Spinocerebellar Ataxia type 1 (nucleotide expansion) *OPL code: SCA1*
- ☐ SCA2 Spinocerebellar Ataxia type 2 (nucleotide expansion) *OPL code: SCA2*
- ☐ SCA3 Spinocerebellar Ataxia type 3 (nucleotide expansion) *OPL code: SCA3*
- ☐ SCA6 Spinocerebellar Ataxia type 6 (nucleotide expansion) *OPL code: SCA6*
- ☐ SCA7 Spinocerebellar Ataxia type 7 (nucleotide expansion) *OPL code: SCA7*
- ☐ SCA12 Spinocerebellar Ataxia type 12 (nucleotide expansion) *OPL code: SCA12*
- ☐ SCA17 Spinocerebellar Ataxia type 17 (nucleotide expansion) OPL code: SCA17
- ☐ Dentatorubral-Pallidoluysian Atrophy (DRPLA) (nucleotide expansion) *OPL code: DRPLA*

● **SINGLE GENE ANALYSIS** *(OPL code: MGDM0)* / **CUSTOM PANEL** (send your request to: polegenetmol@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)

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

● **DNA CONSERVATION** *(OPL code: ADNLD)* **(conservation for 5 years)** ☐

Cerba

MEDICAL PRESCRIPTION FORM

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

MOLECULAR DIAGNOSIS OF A NEUROLOGICAL AND NEURODEVELOPMENTAL DISORDER

CLINICAL INFORMATION

Symptomatic patient ☐ **NO** ☐ **YES: age at symptom onset: ..... years**

Clinical suspicion:

.....

The main clinical signs must be recorded (HPO):

MOST RELEVANT INDICATION ACCORDING TO THE BIOMEDICINE AGENCY'S THESAURUS

Neurodegenerative Diseases

☐ Amyotrophic Lateral Sclerosis or motor neuron disease with or without frontotemporal dementia
☐ Neurodegeneration with Brain Iron Accumulation
☐ Hereditary Spastic Paraplegia
☐ Huntington's Disease
☐ Frontotemporal Dementia
☐ Alzheimer's Disease
☐ Parkinson's Disease
☐ Dementia and Alzheimer's Disease

Neurodevelopmental Disorders

☐ Intellectual Disability (ID) without diagnostic hypothesis
☐ Intellectual Disability (ID) with diagnostic hypothesis
☐ Epilepsies
☐ Intellectual Disability (ID) – Fragile X
☐ Autism Spectrum Disorders

Peripheral Neuropathies

☐ Familial Amyloid Polyneuropathy (TTR gene)
☐ Hereditary Peripheral Neuropathies
☐ Charcot-Marie-Tooth Disease
☐ Hereditary Neuropathies

Central Nervous System

☐ Hereditary Ataxia
☐ Dystonia and Abnormal Movements
☐ Central Nervous System Malformations
☐ Cavernous Angioma
☐ Leukodystrophies and Leukoencephalopathies
☐ Hemiplegic Migraine
☐ Small Vessel Cerebral Disease
☐ Brain Malformations

FAMILY INFORAMTION

Consanguinity	☐ Yes	☐ No
Death in siblings	☐ Yes	☐ No
Affected twinx	☐ 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 .....
Maiden name .....
Address .....
City ..... Country .....
Date of birth: |__|__|__|__|__|__|__|__|
Sampling date: |__|__|__|__|__|__|__|__|

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: |__|__|__|__|__|__|__|__|

Same clinical presentation as the index case patient:
☐ Yes ☐ No *(enclose the clinical description)*