Cerba

FICHE DE PRESCRIPTION MÉDICALE

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

GENETIC DISEASE – MOLECULAR DIAGNOSTIC BY NGS

EXOME AND GENE PANELS
CARDIOGENETICS

SAMPLING

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

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

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

PATIENT

LAST NAME ……

FIRST NAME ……

Birth name ……

Addrese ……

City…… Countr ……

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

Mobile phone : ……

E-mail address ……

EMERGENCY:

☐ Ongoing pregnancy ☐ Prenatal diagnosis ☐ Pediatric resuscitation

PRESCRIER

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)*

☐ 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 EXADP+10003)*

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

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

☐ SOLO (index case only) *(OPL code: EXOME)*

☐ 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 EXADP+10003)*

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

- ☐ Cardiomyopathy Comprehensive Panel (257 genes) *OPL code: IS012*
- ☐ Hypertrophic Cardiomyopathy HAS (18 genes) *OPL code: IS145*
- ☐ Comprehensive Hypertrophic Cardiomyopathy (47 genes) *OPL code: IS013*
- ☐ Dilated Cardiomyopathy (167 genes) *OPL code: IS014*
- ☐ Left Ventricular Noncompaction Cardiomyopathy (46 genes) *OPL code: IS015*
- ☐ Arrhythmogenic Right Ventricular Cardiomyopathy (51 genes) *OPL code: IS016*
- ☐ Transthyretin cardiac amyloidosis (gene *TTR*) *OPL code: IS023*
- ☐ Fabry Disease (gene GLA) OPL code: IS115
- ☐ Lamins-related Cardiomyopathy (gene *FLNA*) *OPL code: IS118*
- ☐ Sudden and Unexplained Death or Survivors of a Cardiac Event (148 genes) *OPL code: IS022*
- ☐ Hereditary Cardiac Disease Panel HAS (48 genes) *OPL code: IS146*
- ☐ Hereditary Cardiac Disease Comprehensive Panel (660 genes) *OPL code: IS011*
- ☐ Arrhythmia Comprehensive Pane (110 genes) *OPL code: IS017*
- ☐ Catecholaminergic Polymorphic Ventricular Tachycardia (13 genes) *OPL code: IS018*
- ☐ Cardiac conduction disorder (36 genes) *OPL code: IS119*
- ☐ Idiopathic ventricular fibrillation (gene *RYR2*) *OPL code: IS120*
- ☐ Long QT Syndrome (38 genes) *OPL code: IS019*
- ☐ Short QT Syndrome (7 genes) *OPL code: IS021*
- ☐ Brugada Syndrome (20 genes) *OPL code: IS020*
- ☐ Jervell and lange-nielsen syndrome (7 genes) *OPL code: IS121*
- ☐ Congenital heart disease (411 genes) *OPL code: IS024*
- ☐ Other panel* :……

Liste des genes sur demande (polegenetmol@lab-cerba.com)

● **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)**

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

● **DNA CONSERVATION** *(OPL code: ADNLD)* **(conservation for 5 years HN200)** ☐

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MEDICAL PRESCRIPTION FORM

GENETIC DISEASE – MOLECULAR DIAGNOSTIC BY NGS

EXOME AND GENE PANELS

CARDIOGENETICS

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

- ☐ Arrhythmogenic right ventricular cardiomyopathy/Left ventricle/BiV (CMP/TR)
- ☐ Cardiomyopathy with left ventricular non-compaction (CMP)
- ☐ Dilated cardiomyopathy (CMP)
- ☐ Hypertrophic cardiomyopathy (CMP)
- ☐ Cardiomyopathy related to ATTR amyloidosis (CMP)
- ☐ Restrictive cardiomyopathy (CMP)
- ☐ Fabry disease (CMP)
- ☐ Laminopathy (CMP)
- ☐ Congenital heart disease
- ☐ Supraventricular arrhythmias (TR)
- ☐ Catecholaminergic polymorphic ventricular tachycardia (TR)
- ☐ Idiopathic ventricular fibrillation (TR)
- ☐ Cardiac conduction disorders (TR)
- ☐ Short QT syndrome (TR)
- ☐ Brugada syndrome (TR)
- ☐ Jervell and Lange-Nielsen syndrome (TR)
- ☐ Long QT syndrome (TR)
- ☐ Sudden death (CM/TR)

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	**FATHER OF THE PATIENT** 2 x 5-mL EDTA tubes of whole blood
LAST NAME ……	LAST NAME ……
FIRST NAME ……	FIRST NAME ……
Birth name ……	
Address ……	Address ……
City …… Country ……	City …… Country ……
Date of birth: |_|_|_|_|_|_|_|_|	Date of birth: |_|_|_|_|_|_|_|_|
Sampling date: |_|_|_|_|_|_|_|_|	Sampling date: |_|_|_|_|_|_|_|_|
Same clinical presentation as the index case patient: ☐ Yes ☐ No *(enclose the clinical description)*	Same clinical presentation as the index case patient: ☐ Yes ☐ No *(enclose the clinical description)*