Cerba

MEDICAL PRESCRIPTION FORM

Laboratoire Cerba
Customer relation service
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Email : intgb@lab-cerba.com

EXOME AND NGS PANELS
GENETIC TESTING IN NEPHROLOGY

SAMPLING

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

DIAGNOSTIC POST-NATAL : ☐ EDTA whole blood (0.5 ml to 5 ml)) ☐ DNA Extract

PATIENT	PRESCRIBER
LAST NAME …………………………………………………………………………	**Numéro RPPS (obligatoire)** : ………………………………………………
FIRST NAME …………………………………………………………………………	Cachet obligatoire
Birth name …………………………………………………………………………	
Address …………………………………………………………………………	
City ………………… Country …………………………………………………	
Date of birth: |_|_||_|_||_|_|_|_|	
N°SS : |_||_|_||_|_||_|_||_|_|_||_|_|_||_|_|_||_|_|	Email address: ……………………………………………………………
EMERGENCY:	**Signature** :
☐ Ongoing pregnancy ☐ Pediatric resuscitation	

REQUESTED TEST
IN CASE OF EMERGENCY A TRIO ANALYSIS IS IMPERATIVE
(One form per sample if request for a TRIO analysis)

- **EXOME ANALYSIS (WES)** (SNV/DELINS and CNV) (≈ 22,000 genes+ ≈ 12,000 No-coding variants (intronic and promoter regions)

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

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

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

- **NGS PANEL ANALYSIS** (SNV/DELINS and CNV) Gene list on request (polegenetmol@lab-cerba.com)

☐ SOLO (only in the index case)

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

☐ TRIO (index case AND its 2 parents)

- ☐ Renal Tubular Acidosis (9 genes) *OPL code: IS094*
- ☐ Tubulopathies (86 genes) *OPL code: IS109*
- ☐ Nephrolithiasis/Nephrocalcinosis (60 genes) *OPL code: IS110*
- ☐ Primary hyperoxaluria (3 genes) *OPL code: IS059*
- ☐ Hypertension Pseudoaldosteronism (22 genes) *OPL code: IS111*
- ☐ Familial polycystic kidney disease (58 genes) *OPL code: IS037*
- ☐ Hemolytic uremic syndrome (23 genes) *OPL code: IS053*
- ☐ Renal amyloidosis (5 genes) *OPL code: IS112*
- ☐ Nephronophtisis/Renaliliopathy (86 genes) *OPL code: IS077*
- ☐ Nephrotic syndrome/proteinuria (112 genes) *OPL code: IS078*
- ☐ Developmental abnormalities of the kidney and urinary tract (CAKUT) (170 genes) *OPL code: IS113*
- ☐ End-stage renal disease in young people (253 genes) *OPL code: IS114*
- ☐ KIDNEY DISEASES Comprehensive Panel (867 genes) *OPL code: IS093*
- ☐ Alport syndrome/hematuria (6 genes) *OPL code: IS003*
- ☐ Bardet-Biedl syndrome (39 genes) *OPL code: IS006*
- ☐ Syndrome de Bartter/Gitelman (32 genes) *OPL code: IS007*
- ☐ Branchio-Oto-Renal Syndrome (4 genes) *OPL code: IS009*
- ☐ Joubert and Meckel syndrome (52 genes) *OPL code: IS063*
- ☐ Fabry disease (gene *GLA*) *OPL code: IS115*
- ☐ Acute intermittent porphyria (gene *HMBS*) *OPL code: IS116*
- ☐ Cystinosis (gene *CTNS*) *OPL code: IS117*

- **APOL1 – ASSOCIATED NEPHROPATHY (APOL1 GENOTYPING)** (OPL code: *APOL1*)

- **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)* **(5-year shelf life HN200)** ☐

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CLINICAL INFORMATION

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

Diagnostic hypothesis:

.....

Main clinical signs must be recorded (HPO):

MOST RELEVANT INDICATION ACCORDING TO THE THESAURUS OF THE BIOMEDICINE AGENCY

☐ Kidney and urinary tract development abnormalities (CAKUT)
☐ Ciliopathy with renal involvement (except autosomal dominant polycystic fibrosis)
☐ Complement susceptibility factors for atypical HUS
☐ Lithiasis or nephrocalcinosis
☐ Autosomal dominant cystic kidney disease including autosomal dominant polycystic fibrosis
☐ Familial hematuric nephropathy
☐ Indeterminate nephropathy with renal failure
☐ Autosomal dominant tubulointerstitial nephropathy (ADTKD)
☐ Glomerulopathy including proteinuria and cortico-resistant nephrotic syndrome
☐ Tubulopathie

FAMILY INFORMATION

Consanguinity	☐ Yes	☐ No
Death in siblings	☐ Yes	☐ No
Affected twins	☐ Yes	☐ No

FAMILY TREE

☐ Man
☐ Woman
◇ Sex unknown
■ □ ◆ Individual of unknown sex
☐ □ ◇ Healthy subjects

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: |_|_|/|_|_|/|_|_|_|_|

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

FTAHER 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 index patient:
☐ Yes ☐ No *(enclose the clinical description)*