INFORMATION FOR GENETIC CHARACTERS INVESTIGATION

Developmental disorder and genetic diseases

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AUTHORIZED LABORATORY FOR PERFORMING GENETIC CHARACTERS INVESTIGATION IN HUMAN

PATIENT	PRESCRIBING PHYSICIAN
Name..... First name..... Birth date ..... Address.....	Signature :

FAMILY INFORMATION	CLINICAL INFORMATION
SPOUSE Name.....First name..... **FATHER** Name..... First name..... **MOTHER** Name..... First name.....	☐ Pregnancy (since) ⌊_⌋_⌋_⌋_⌋_⌋ ☐ Ongoing prenatal diagnostic

CYTOGENETIC & MOLECULAR CYTOGENETIC (mandatory clinical information)

☐ Total blood Number of samples : ..... ☐ EDTA ☐ Heparinized
☐ Tissue (in culture medium) : specify..... .....

REQUIRED TEST

☐ Constitutional karyotype from patient (+ 8 days old) blood (Heparin tube)
☐ Chromosomal micro-array (CMA) (EDTA tube)
☐ Constitutional karyotype from newborn (0 to 8 days) blood (Heparin tube)
☐ (pan-) telomeric study (MLPA, EDTA tube)
☐ Microdeletion syndrome screen (FISH technique):
o Wolf-Hirschhorn (4p-) o Cri du Chat (5p-) o Willi-Prader o Angelman o Williams-Beuren
o Smith-Magenis o Miller-Diecker o DiGeorge o Other.....

☐ Uniparental disomy (specify chromosome) : .....
☐ Other (specify) : .....

INDICATION

Intellectual disability/ Malformations

☐ (12) Intellectual disability
☐ (32) Autism spectrum
☐ (14) Dysmorphic features
☐ (20) Obesity with intellectual disability
☐ (33) Delayed language
☐ (35) Behavioral disorder
☐ (46) Congenital abnormality (precise) :.....
☐ (23) Other (specify) : .....

☐ (34) Pervasive developmental disorder
☐ (36) Developmental delay
☐ (30) Hypotonia
☐ (40) Epilepsy
☐ (40) Suspicion of fragile X

☐ (22) Suspicion of trisomy 21
Maternal serum markers
(child under 1):
☐ Yes :
☐ 1st T ☐ 2nd T
☐ 2nd T integrated ☐ Indetermined
Result 1/.....
☐ No

Gonosomic abnomalitis suspicion

☐ (02) Klinefelter Syndrome
☐ (05) Primary amenorrhea
☐ (06) Secondary amenorrhea
☐ (18) Stature/weight retardation

☐ (27) Turner Syndrome
☐ (07) Early menopause
☐ (17) Ambiguous genitalia / genital malformations
☐ (19) Delayed puberty

☐ (31) Premature ovarian failure
☐ (28) Gender dysphoria
☐ (21) Obesity without intellectual disability
☐ (03) Gynecomastia

Reproductive disorders

☐ (01) Azoospermia
☐ (01) AVD
☐ (11) Multiple miscarriage (number) : .....

☐ (01) Severe oligospermia
☐ (08) Check up before IVF/ICSI/gamete donation

☐ (01) Oligo-astheno-teratospermia OATS
☐ (10) Non-labeled infertility

Familial investigation chromosomal abnormality (send index case results or name, address and phone number of the lab which performed the karyotype)

☐ (25) Familial study (1st degree related)
☐ (26) Prenatal diagnosis during

☐ (29) Familial study (non 1st degree related)

MOLECULAR GENETIC (mandatory clinical information)	
□ Total blood Number of samples: ……..	□ EDTA
□ Tissue (in culture medium) : specify…… ……	
□ Urine (morning urine)	

REQUIRED TEST	**INDICATION**
□ Frequent mutations screen for CFTR gene (+/-IVS8 (T)(TG) splicing variant +/- rare mutations) (reflex test) □ Y-chromosome microdeletions □ *SRY* gene screen	□ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/CFTRGB.pdf □Investigation of male infertility □ Azoospermia □ Severe oligospermia □ OATS □ Other *(specify)* :…… □ Sexual ambiguity □ Other *(specify)* :……
□ *FGFR3* Achondroplasia □ *FGFR3* Hypochondroplasia □ *FGFR3* Thanatophore dysplasia □ *FGFR2* Apert syndrome □ *SHOX* Léri-Weill and Langer syndromes, idiopathic small size □ *PTPN11* Noonan syndrome / Leopard syndrome	□ Clinical suspicion □ Family study □ Other *(specify)* :…… □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/SHOXGB.pdf □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/NOONGB.pdf
□ *FMR1* gene study (fragile X syndrome) □ Angelman syndrome □ Willi-Prader syndrome □ *MECP2* gene study (Rett syndrome)	□ Clinical suspicion □ Family study □ Other *(specify)* :…… □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/RETTGB.pdf
□ *DMPK* Steinert myotonic dystrophy* □ *SMN1* Spinal muscular atrophy (diagnostic) □ *SMN1* Spinal muscular atrophy (heterozygote)*	□ Clinical suspicion □ Family study □ Other *(specify)* :……
□ *GJB6* Connexin 30 □ Mitochondrial deafness □ Mitochondrial deafness / diabetes □ *GJB2* Connexin 26	□ Clinical suspicion □ Family study □ Other *(specify)* :…… □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/CONNGB.pdf
□ Mitochondrial cytopathy MERRF □ Mitochondrial cytopathy MELAS □ Mitochondrial cytopathy es NARP □ Leber's optical atrophy LHON □ Mitochondrial deafness □ Mitochondrial deafness / diabetes	□ Clinical suspicion □ Family study □ Other *(specify)* :……
□ *HEXA* Tay-Sachs disease □ *ASPA* Canavan disease □ *IKBKAP* Familial dysautonomy □ *AAT* alpha-1 antitrypsin genotype □ *UGT1A1* Gilbert disease □ Fabry disease (alpha-galactosidase assay) □ *MEFV* Familial Mediterranean Fever and others HRF □ *F8/F9* A & B hemophilia □ *HBB/HBA1/HBA2* Sickle-cell disease and other Hemoglobin diseases	□ Clinical suspicion □ Biological suspicion □ Heterozygote screening □ Personal familial history □ Spouse familial history □ Without previous history □ Other *(specify)* :…… □ Molecular study of an index case □ Other *(specify)* :…… □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/GFMFGB.pdf □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/HEABGB.pdf □ **Send specific clinical information sheet** https://www.lab-cerba.com/files/live/sites/Cerba/files/documents/EN/HEMOGB.pdf