Hôpitaux Universitaires Genève

DIAGNOSTIQUE / Service de Médecine Génétique
Centre d'accueil des prélèvements (CAP)
Bâtiment des Laboratoires (BATLab), local 8D-0-850.1
4 rue Gabrielle-Perret-Gentil, 1211 Genève 14

Genomic and Molecular Diagnostics Laboratory
Accredited since 2003, formerly STS 0382

DIAGMOL

http://www.hug-ge.ch/feuilles-de-demande

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Lab direct or results: Phone/FAX: +41 (0) 22 37 21 826 / 21 860
Sample Entrance Center (CAP) : Phone +41 (0) 22 37 21 800

Mr. ❑ Mrs. ❑ (IN UPPER CASE, please)

Name:……

Maiden Name:……..

First name :……..

Date of birth : ..... / ..... / ……

Legal representative (for minors) : ❑father ❑mother

Name/first name :……

Street/N°:……

Town, ZIPCODE :……

Hospitalisation Unit: …… Physician :……

N° EdS : ……

Invoice address: ❑Patient ❑Prescriptor ❑Insurance

Type of case : ❑Disease ❑AI ❑Accident ❑Pregnancy

N° AVS (Mandatory for AI) : ……

Insurance : …… **Insured N :** ……

PHYSICIAN

PHYSICIAN *(NAME/First name - Street/N°- Town, ZIPCODE - Phone/FAX. IN UPPER CASES, PLEASE)*

COPY TO OTHER PHYSICIAN *(NAME/First name - Street/N°- Town, ZIPCODE - Phone/FAX. IN UPPER CASES, PLEASE)*

« The laboratory is granted permission by the Physician/Patient to transmit copies of the report to other physicians»

❒ **Opposition of the patient to the registration of this request results in the electronic patient record (DPI) of the HUG**

If the patient belongs to a family already known to the laboratory, please indicate index case NAME:

CLINICAL INFORMATIONS given by the physician:

Ethnic origins | Father | Mother

Currently pregnant ☐ **Date of last menses** **Number of weeks of amenorrhea**

SAMPLE(S)

*Most of our tests work from 4 ml of blood in **EDTA** (children <2 ans : 1 ml : ok) or from **purified DNA** (some exceptions apply for some tests) Please contact us for any other type of sample.*

On every and single tube. **Mandatory** !

NAME First name
Date of birth

Sampling date : **Time (optional) :** **Sample Number :**

❑ Blood (tube EDTA)

❑ Saliva (sampled only in tube Oragene-DNA)

❑ Other type of sample
Please indicate type :

❑ DNA from external source
Reference :

❑ DNA already in bank at our laboratory
Reference number:
(if known)

Prenatal
❑ Amniotic fluid
❑ Chorionic villi
❑Fetal tissue
❑Other

Réservé au laboratoire

Tissu	O sang/EDTA	O ADN	O ADN déjà en banque
	O Liq. Amnio.	O CVS	O Tissus fixés
	O Liq. Amnio.	O CVS	O Tissus fixés

Quantité, Remarques :

TEST LIST : SEE OVER (PAGES 3 ET 4)

NAME, First name :
(CAPITAL LETTERS, please)

Laboratory only
DM-

PHYSICIAN'S SIGNATURE AND INFORMED CONSENT

PHYSICIAN *(NAME/First name – Street/N°- Town, ZIPCODE - Phone/FAX) :*

** I certify that the concerned person (patient, legal representative) has received genetic counseling according to the law on human genetic analysis (LAGH) on the various aspects of the described genetic analysis in the form "patient information". This person has given its consent **(in writing for prenatal, presymptomatic or family planning analyses)** and had enough time to ask questions and make its decision.*

Date and Physician's signature **MANDATORY***

The patient has given his informed consent for the checked analyses that are listed at the pages 3-4, to be done on the sample specified at the first page.

*The following questions marked by a star * must be checked (MANDATORY !).*

Decision of the patient regarding the storage and use of his/her remaining biological sample(s) and raw analytical data : **mandatory*

- He/she agrees that the remaining biological material and raw analytical data will be stored for possible further analyses. His/her informed consent will be necessary for any further additional analyses. * ☐YES ☐NO
  In case of a negative answer the remaining biological sample will be destroyed after the analysis.

- He/she agrees that his/her biological sample and raw analytical data are used anonymously for quality testing. * ☐YES ☐NO

* **MANDATORY ONLY FOR ANALYSES INVOLVING HIGH THROUGHPUT SEQUENCING OF WHOLE EXOME (SEE PAGE 4)**

Decision of the patient regarding the transmission of results not directly related to the testing requested (so called "incidental findings") **mandatory*

He/she wishes to be informed about genetic results belonging to the following categories :

- Carrier of a disorder for which preventive and/or therapeutic measures are available* : ☐YES ☐NO

Person incapable of discernment: ☐YES ☐NO

The following questions do NOT apply for persons incapable of discernment

- Carrier of a disorder for which no preventive / therapeutic measures are yet available* : ☐YES ☐NO
- Healthy carrier of a recessive disorder which could concern the following generation or other family members* : ☐YES ☐NO
- Other decisions : _____

OPTIONAL

The use of his/her sample and data for research purposes.

Should he/she agree in principle to participate in research studies you could indicate this below. Should this be the case he/she would be contacted at a later stage with details concerning the research project(s). A positive answer below is **not yet consent** for the participation in any actual research projects**.**

- In principle, he/she agrees that his/her biological sample and data could be used for research purposes ☐YES ☐NO

NAME, First name :
(CAPITAL LETTERS, please)

Laboratory only
DM-

REQUESTED ANALYSIS / ANALYSES

NB: IF REQUEST IS FOR A HIGH THROUGHPUT SEQUENCING (GENOME CLINIC), PLEASE GO DIRECTLY TO PAGE 4.

✖ Test not included in the Swiss federal list of laboratory tests (OFSP, BAG, FOPH). The out-of-list tests are not automatically reimbursed by Swiss health insurances. na Test not accredited; @ Please contact us in advance . The rates applied are those of the list of analyzes of the OFSP / BAG. The laboratory reserves the right to select the most appropriate technique (traditional or high throughput sequencing, cf. page 4) based on efficiency and cost effectiveness. cl Depending on the choice of technique, the analysis may or may not be accredited.

General tests
- ❒ Amyloidosis (familial, *TTR)*
- ❒ AS, Angelman syndrome na
- ❒ APECED (*AIRE)*
- ❒ Beckwith-Wiedemann (BWS) na
- ❒ BPES (*FOXL2)* ✖
- ❒ CMM2 Cutan. Malign. Melanoma (*CDKN2A*) ✖na
- ❒ *EGFR* mutations (T790M and others) on ctDNA (only in Streck BCT or PAXgene DNA tubes) ✖
- ❒ FG (Keller syndrome, *MED12)* ✖na
- ❒ HBLRG, Gilbert syndrome (*UGT1A1*) ✖
- ❒ HDGC, Her. Diff. Gastric Cancer (*CDH1*) ✖na
- ❒ HED, Hypohidrotic Ectodermal Dysplasia (*EDA)* ✖
- ❒ HFE-HH, Hered. Hemochromatosis (*HFE*)
- ❒ HSCR, Hirschsprung (*RET)* ✖na
- ❒ XLI, Ichtyosis, X-linked type (*STS*)
- ❒ PFIC3, Intrahepatic Cholestasis (*ABCB4*) ✖cl
- ❒ SMAX1, Kennedy *(*SBMA*, AR)* na
- ❒ KNO1, Knobloch*, (COL18A1)* na
- ❒ Lactose intolerance (*LCT*) ✖
- ❒ LWD, Leri-Weill (*SHOX*)
- ❒ LFS, Li Fraumeni, (*TP53)*
- ❒ Marfan (*FBN1)*
- ❒ NF1, Neurofibromatosis type I (*NF1*) na
- ❒ Non-invasive prenatal diagnostic of monogenic diseases (*contact us in advance)* ✖
- ❒ PJS, Peutz-Jeghers (*STK11)* na
- ❒ PTEN Hamartoma Tumor syndrome (PHTS, Cowden, Hamartomas, BRRS, Proteus, *PTEN*)
- ❒ PWS, Prader-Willi na
- ❒ Rendu-Osler-Weber (ROW) ✖cl @
- ❒ RETT syndrome (*MECP2)*
- ❒ RSS, Russell-Silver syndrome (11p15) na
- ❒ Sickle cell anemia (Drepanocytosis, *HBB*)
- ❒ UPD, Uniparental Disomy, **Chr_____** ✖
- ❒ VWF, all types, ✖na @
- ❒ WAGR, Wilms tumor (*WT1*) ✖na

Alpha-1-antitrypsin deficiency (A1AT)
- ❒ Genotyping PI*S/Z
- ❒ Full sequencing of *SERPINA1*

Ashkenazi mutations (rare disease carrier)
- ❒ Full screening ✖ or
- ❒ *CFTR* ❒ Fragile-X
- ❒ Tay-Sachs+ FD+Fanconi+Canavan ✖
- ❒ von Gierke+Bloom+Niemann-Pick+ML-IV ✖

Individual prices available upon request

Ataxias
- ❒ Full screening
- ❒ SCA1 na ❒ SCA2 na ❒ SCA3 na
- ❒ SCA6 na ❒ SCA7 na ❒ SCA17 na
- ❒ Friedreich ❒ DRPLA na ❒ *FXTAS* na

Cardiac Arrhythmias (Channelopathies, CCP) ✖
- ❒ *SCN5A* gene (Brugada) cl
- ❒ *KCNQ1* gene (QT-long) cl
- ❒ *KCNH2* gene (QT-long) cl
- ❒ *KCNE1, KCNE2, KCNJ2 genes* cl

Whole Exome Sequencing and Targeted Gene Panel Analysis : see next page (page 4)

Cardiomyopathies (HCM, DCM, NC, CMR,…) ✖
Whole Exome Sequencing and Targeted Gene Panel Analysis: *see next page (page 4)*

Charcot-Marie-Tooth (CMT)
- ❒ Duplication CMT1A
- ❒ *PMP22* gene sequencing (CMT1A)
- ❒ *MPZ* gene sequencing (CMT1B)
- ❒ *GJB1* gene sequencing (CMTX)

Chromosomal Microdeletions ✖
- ❒ 22q11, MLPA
- ❒ Screening, recurrent microdeletions, MLPA

Cystic Fibrosis (CF, *CFTR*)
Please indicate the ethnic origins of the patient at page 1 (for residual risk calculation)
- ❒ Screening frequent mutations (with / without IVS8 5T, *CFTR*-related disorders)
- ❒ Full *CFTR* analysis (sequencing+ del/dup by MLPA) na
- ❒ Hyperechogenic fetal bowel (frequent mutations *CFTR* in parents)
- ❒ Hyperechogenic fetal bowel (frequent mutations *CFTR* +del/dup by MLPA in parents)

Deafness
- ❒ DFNB1, congenital (locus DFNB1) ✖
- ❒ Mitochondrial mutations ✖na

Endocrine Neoplasias, Pheochromocytoma, Paraganglioma (MEN, PCC, PGL)
- ❒ MEN1, Multi. Endoc. Neopl. type I (*MEN1)*
- ❒ MEN2, Multi. Endoc. Neopl. type II (*RET*)
- ❒ PGL/PCC, Paraganglioma/Pheochromocytoma: Full sequencing (+MLPA) or
  - ❒ *SDHB* gene ❒ *RET gene*
  - ❒ *SDHC* gene cl ❒ *SDHD* gene

Individual prices available upon request
- ❒ Von Hippel Lindau (*VHL*)

Familial Pneumothorax (Birt-Hogg-Dubé, BHD) ✖na
- ❒ Frequent mutation, *FLCN*, exon 11
- ❒ *FLCN* full gene analysis

Familial Adenomatous Polyposis (FAP)
- ❒ Full Screening *APC* + *MUTYH* or
- ❒ *APC* gene
- ❒ *MAP* (*MUTYH, ex. 7,13*) na

FGFR3 (syndromes linked to)
- ❒ Achondroplasia
- ❒ Craniosynostosis or Muenke
- ❒ Hypochondroplasia
- ❒ Thanatophoric dysplasia, types I, II
- ❒ SADDAN

Fibrinopathies ✖
- ❒ Afibrinogenemia (*FGA, FGB, FGG*)
- ❒ Dysfibrinogenemia (*FGA, FGG*)
- ❒ Hypofibrinogenemia (*FGA, FGG*)

Fragile X *(FRAXA, FMR1)* na
- ❒ Diagnostic
- ❒ Carrier testing
- ❒ Premature ovarian failure (POI)

Genetic sex✖
- ❒ Genetic sex determination
- ❒ *SRY* search in a Turner
- ❒ *SRY* sequencing

Hemophilias
- ❒ HA, inversions *F8* (IVS22, IVS1) ✖
- ❒ HA, *F8,* complete analysis cl
- ❒ HB, *F9,* complete analysis

Huntington disease (HD, *HTT*)
- ❒ Diagnostic
- ❒ Presymptomatic ***(2 tubes please)***

Hereditary Periodic Fevers (HRF) ✖
- ❒ Full Sequencing (8 genes)
- ❒ Full Screening Frequent Mutations (4 genes)
- ❒ FMF, *MEFV* gene
- ❒ FMF, *MEFV* gene (complete sequencing)
- ❒ CAPS, *NLRP3* gene
- ❒ HIDS, *MVK* gene
- ❒ TRAPS, *TNFRSF1A* gene

Whole Exome Sequencing and Targeted Gene Panel Analysis: *see next page (page 4)*

HNPP (tomaculous neuropathy)
- ❒ Deletion CMT1A
- ❒ *PMP22* gene sequencing

Lynch syndrome (HNPCC)
Full screening *by NGS see next page (page 4)* or
- ❒ *MLH1+PMS2* genes
- ❒ *MSH2+MSH6* genes
- ❒ MSI (on tumor biopsy)
- ❒ *BRAF1 V600E* (on tumor biopsy) na

Male infertility
- ❒ *CFTR*+5T (frequent mutations)
- ❒ Y chromosome microdeletions (DAZ)

Mitochondriopathies (ADNmt)
- ❒ Leber optic neuropathy (LHON)
- ❒ Cytopathy MELAS, MERRF, NARP
- ❒ Deletions/Dup ***(muscle biopsy only)*** ✖
- ❒ Chromosome Mit sequencing

Monogenic Diabetes (MODY, NDM) ✖
Whole Exome Sequencing and Targeted Gene Panel Analysis: *see next page (page 4)* Or
- ❒ *HNF4A* gene (MODY 1) cl
- ❒ *GCK* gene (MODY 2) cl
- ❒ *HNF1A* gene (MODY 3) cl
- ❒ *PDX1* gene (MODY 4) cl
- ❒ *HNF1B* gene (MODY 5) cl
- ❒ *INS* gene (MODY 10) cl
- ❒ *KCNJ11* gene cl

Neurological and Neuromuscular
- ❒ SMA, Spinal Musc. Amyotrophy (*SMN1*)
- ❒ CADASIL (*NOTCH3)* ✖
- ❒ C9orf72 (ALS, FTD) ✖
- ❒ DOPA-responsive dystonia (*GCH1*) ✖na
- ❒ Dravet syndrome (*SCN1A)* ✖na
- ❒ EPM1, Unverricht-Lundborg (*CSTB*) ✖ Southern+sequencing
- ❒ FSHD1, Facio-Scapulo-Humeral Dystrophy, type1✖ na ***(only from fresh EDTA blood)***
- ❒ GLUT1 (*SLC2A1*) ✖na
- ❒ SPAST, Hered. Spastic Paraparesis (*SPG4*)
- ❒ STARTLE (Hyperekplexia, *GLRA1*) ✖na
- ❒ DMD, BMD, Muscular Dystroph Duchenne/Becker (*DMD*), deletions
- ❒ DM1, Myotonic Dystrophy of Steinert (*DMPK*)
- ❒ OPMD, Oculopharyngeal Muscular Dystrophy (*PABPN1*) ✖na
- ❒ TOR1A, Torsion Dystonia (*DYT1*) ✖na

Pancreatitis
- ❒ *CFTR*+ IVS8 5T (frequent mutations)
- ❒ *SPINK* frequent mutation
- ❒ *PRSS1* frequent mutations ✖

Primary Ciliary Dyskinesia (PCD) ✖
Whole Exome Sequencing and Targeted Gene Panel Analysis: *see next page (page 4)*

Waardenburg (WS) ✖
- ❒ Types I and III *(PAX3* gene*)*
- ❒ Type II *(MITF* gene) na

Miscelleanous (cf. additional informations)
- ❒ **DNA** Extraction + Banking
- ❒ Circulating cell-free DNA Extraction (only in Streck-Cell free BCT tubes)
- ❒ *Out-of-list (OFAS) gene* na✖*per exon*
- ❒ Specific/ Known familial mutation ✖
- ❒ mRNA analysis ✖na @ (blood only in PAXgene blood RNA tubes)
- ❒ Exclusion of maternal contamination in fetal sample *(amnio-, choriocentesis)* ✖
- ❒ Transfer of DNA to an external laboratory (please provide specifics below) and complete the ECA forms for internal requests.
- ❒ Transfer of DATA of NGS to an external laboratory (please provide specifics below), complete the DATA exchange form and complete the consent level 3.

Please indicate here any additional helpful information, other specific tests, desired order of analyses (for multiple tests), gene panel ….

NAME, First name :
(CAPITAL LETTERS, please)

Laboratory only
DM-

REQUESTED ANALYSIS / ANALYSES

HIGH THROUGHPUT SEQUENCING OF TARGETED OR WHOLE EXOME AND BIOINFORMATIC ANALYSIS (GENOME CLINIC)

NB: IF THE REQUEST CONCERNS A CLASSICAL ANALYSIS, WITHOUT HIGH THROUGHPUT SEQUENCING, SEE PAGE 3

✖ Test not included in the Swiss federal list of laboratory tests (OFSP, BAG, FOPH). The out-of-list tests are not automatically reimbursed by Swiss health insurances.
[na] Test not accredited; @ Please contact us in advance (availability, TAT,…). The rates applied are those of the list of analyzes of the OFSP / BAG. The laboratory reserves the right to select the most appropriate technique (high throughput sequencing or traditional, cf. page 3) based on efficiency and cost effectiveness.

❒ **DNA extraction and banking**

HIGH THROUGHPUT SEQUENCING FOLLOWED BY BIOINFORMATIC ANALYSIS OF 1 TO 10 GENES @

Prescription by a physician with a federal postgraduate FMH diploma in medical genetics or related to the examined pathology, according to the Swiss federal list of laboratory tests (Chapter 2.Genetics na 2.2.2.Moleculare genetics analyses).

❒ **Cardiac Channelopathies (Arrhythmias, CCP)** ✖
❒ **Cardiomyopathies** ✖
❒ **Diabetes, monogenic (MODY, NDM)** ✖
❒ **Primary Ciliary Dyskinesia (PCD)** ✖
❒ **FGF receptor-associated dysplasias**
❒ **Duchenne and Becker dystrophinopathies and muscular dystrophies (protein disorders associate with dystrophin)**
❒ **Hereditary periodic fevers** ✖
❒ **Wilson's disease**
❒ **Hereditary neoplasia**
❒ **Lynch syndrome (HNPCC)**
❒ **Neurofibromatosis type I**
❒ **Growth disorder syndromes**
(Beckwith-Wiedemann, Silver-Russel, Sotos, etc)

❒ **Other disease (please specify below the genes to analyze and the position in the Swiss list of laboratory tests)**

❒ **Other orphan diseases** ✖ **(please specify below the genes to analyze)**
An "Orphan disease" reimbursement request must be filled by a physician with a federal postgraduate FMH diploma

HIGH THROUGHPUT SEQUENCING FOLLOWED BY A BIOINFORMATIC ANALYSIS OF MORE THAN 10 GENES @

Prescription only by a physician with a federal postgraduate FMH diploma in medical genetics, according to the Swiss federal list of laboratory tests.

Mitochondriopathies
❒ 11-100 genes
❒ > 100 genes
Cardiac Channelopathies (Arrhythmias, CCP) ✖
❒ 11-100 genes
Cardiomyopathies ✖***na***
❒ 11-100 genes
Primary Ciliary Dyskinesia (PCD) ✖
❒ 11-100 genes
Diabetes, monogenic (MODY, NDM) ✖
❒ 11-100 genes
Ehlers-Danlos
❒ 11-100 genes
Epilepsy ✖
❒ 11-100 genes
❒ > 100 genes
Diseases related to coagulation, blood and immune system disorders
❒ 11-100 genes
❒ > 100 genes
Neuromuscular et neurodegenerative diseases
❒ 11-100 genes
❒ > 100 genes
Diseases related to skin, connective tissue or bones
❒ 11-100 genes
❒ > 100 genes
Metabolic and endocrine diseases
❒ 11-100 genes
❒ > 100 genes
Ophtalmologic diseases
❒ 11-100 genes
❒ > 100 genes

Diseases related to urogenital system, fertility / sterility
❒ 11-100 genes
❒ > 100 genes
Hereditary neoplasia
❒ 11-100 genes
❒ > 100 genes
Sensorimotor neuropathies
(CMT, HNPP, amyloid polyneuropathy)
❒ 11-100 genes
Kallman syndrome
❒ 11-100 gènes
Marfan syndrome and other thoracic aorta syndromes
❒ 11-100 genes
Mendelian syndrome with growth disorder
❒ 11-100 genes
❒ > 100 genes
Developmental disorders ✖
❒ 11-100 genes
❒ > 100 genes

Other diseases (please specify below the genes to analyze and the position in the Swiss list of laboratory tests)
❒ 11-100 genes
❒ > 100 genes

Other orphan diseases ✖ **(please specify below the genes to analyze)**
An "Orphan disease" reimbursement request must be filled by a physician with a federal postgraduate FMH diploma
❒ 11-100 genes
❒ > 100 genes

ADDITIONAL ANALYSES @

Additional bioinformatic analysis ❒ 1-*10 genes* ❒ *11-100 genes* ❒ *more than 100 genes*
Other additional analyses ❒ Sanger sequencing ❒ MLPA ❒ Other ([na]*, depending which analysis*) : _____
Comments:_____

INFORMATIONS ABOUT BIOINFORMATIC ANALYSES Gene panels: http://www.hug-ge.ch/medecine-genetique/liste-panels-genes
Gene panel to analyze *(please contact us in advance):*
_____
*(Please specify the investigated pathology, the number of genes and the requested gene panel (if available) or else provide **your gene list in an Excel file**)*

SEARCH FOR VARIANTS IN THE PARENTS

In case of request of search of variants in the parents, please send us for each of them the sample and a request of DNA extraction and banking.

❒ Consanguineous parents
Precisions/comments :_____
Father: Last name :_____ First name :_____ Date of birth :_____ Sample available : ❒ Yes ❒ No ❒ Will be sent
Mother: Last name :_____ First name :_____ Date of birth :_____ Sample available : ❒ Yes ❒ No ❒ Will be sent

Complementary information /comments: