

**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 Prescripitor 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 | <input type="checkbox"/> sang/EDTA   | <input type="checkbox"/> ADN | <input type="checkbox"/> ADN déjà en banque |
|       | <input type="checkbox"/> Liq. Amnio. | <input type="checkbox"/> CVS | <input type="checkbox"/> Tissus fixés       |
|       | <input type="checkbox"/> Liq. Amnio. | <input type="checkbox"/> CVS | <input type="checkbox"/> Tissus fixés       |

Quantité, Remarques :

**TEST LIST : SEE OVER (PAGES 3 ET 4)**

NAME, First name :  
(CAPITAL LETTERS, please)

Laboratory only

DM-

Report in English (default: in French)

## PHYSICIAN'S SIGNATURE AND INFORMED CONSENT

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

*\* By signing here, the physician confirms having informed the patient/the legal representative according to the current legal requirements (LAGH in Switzerland, <http://www.sgm.ch>) (including on the cost of tests that are not covered by health insurances), that the patient/the legal representative had enough time to ask questions and take his/her decision, and having received the patient's/the legal representative's informed consent.*

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

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

**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 <sup>na</sup>  SCA2 <sup>na</sup>  SCA3 <sup>na</sup>
- SCA6 <sup>na</sup>  SCA7 <sup>na</sup>  SCA17 <sup>na</sup>
- Friedreich  DRPLA <sup>na</sup>  FXTAS <sup>na</sup>

**Cardiac Arrhythmias (Channelopathies, CCP) \***

- SCN5A gene (Brugada) <sup>cl</sup>
- KCNQ1 gene (QT-long) <sup>cl</sup>
- KCNH2 gene (QT-long) <sup>cl</sup>
- KCNE1, KCNE2, KCNJ2 genes <sup>cl</sup>

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) <sup>na</sup>
  - 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 \*<sup>na</sup>

**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 <sup>cl</sup>  SDHD gene
- Individual prices available upon request
- Von Hippel Lindau (VHL)

**Familial Pneumothorax (Birt-Hogg-Dubé, BHD) \*<sup>na</sup>**

- 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) <sup>na</sup>

**FGFR3 (syndromes linked to)**

- Achondroplasia
- Craniosynostosis or Muenke
- Hypochondroplasia
- Thanatophoric dysplasia, types I, II
- SADDAN

**Fibrinopathies \***

- Afibrinogenemia (FGA, FGB, FGG)
- Dysfibrinogenemia (FGA, FGG) 530 -
- Hypofibrinogenemia (FGA, FGG) 530 -

**Fragile X (FRAXA, FMR1) <sup>na</sup>**

- 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 <sup>cl</sup>
- 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) <sup>na</sup>

**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) <sup>cl</sup>
  - GCK gene (MODY 2) <sup>cl</sup>
  - HNF1A gene (MODY 3) <sup>cl</sup>
  - PDX1 gene (MODY 4) <sup>cl</sup>
  - HNF1B gene (MODY 5) <sup>cl</sup>
  - INS gene (MODY 10) <sup>cl</sup>
  - KCNJ11 gene <sup>cl</sup>

**Neurological and Neuromuscular**

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

**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)

**Wardenburg (WS) \***

- Types I and III (PAX3 gene)
- Type II (MITF gene) <sup>na</sup>

**Miscellaneous (cf. additional informations)**

- DNA Extraction + Banking
- Circulating cell-free DNA Extraction <sup>na</sup> (only in *Streck BCT* or *PAXgene blood DNA tubes*)
- Out-of-list (OFAS) gene <sup>na</sup> \*<sub>per exon</sub>
- Specific/ Known familial mutation \*
- 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.

<sup>na</sup> 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.Moleculaire 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 \*<sup>na</sup>**

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

**Ophthalmologic 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 genes

**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 (<sup>na</sup>, 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: