

**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**



**DIAGMOL**

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

Head of Genetic Medicine Division : Prof. Marc ABRAMOWICZ  
Laboratory managers (medical genetics FAMH):  
Dr. J.-L. BLOUIN, Dr. Th. NOUSPIKEL, Dr. M. GUIPPONI  
jean-louis.blouin@hcuge.ch, thierry.nouspikel@hcuge.ch, michel.guipponi@hcuge.ch  
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 Prescriber 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 INFORMATION 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
- 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)**

Report in English (default: in French)

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) :

*\* By signing here, the physician confirms having informed the patient/the legal representative according to the current legal requirements (LAGH in Switzerland, <http://www.sgmg.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.  
All prices are in Swiss Franc (CHF), not including DNA extraction cost (see DNA banking). For prenatal tests (urgent), an additional charge applies (200 CHF). 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.

**General tests**

- Amyloidosis (familial, TTR) 960 CHF
- AS, Angelman syndrome na 533 CHF
- APECED (AIRE) 1605 CHF
- Beckwith-Wiedemann (BWS) na 533 CHF
- BPES (FOXL2) na 530-880 CHF
- CMM2 Cutan. Malign. Melanoma (CDKN2A) na 745-1095 CHF
- EGFR mutations (T790M and others) on ctDNA (only in Streck BCT or PAXgene DNA tubes) na 631 CHF
- FG (Keller syndrome, MED12) na 530 CHF
- HBLRG, Gilbert syndrome (UGT1A1) na 285 CHF
- HDGC, Her. Diff. Gastric Cancer (CDH1) na 315 CHF
- HED, Hypohidrotic Ectodermal Dysplasia (EDA) 1605-1955 CHF
- HFE-HH, Hered. Hemochromatosis (HFE) 186 CHF
- HSCR, Hirschsprung (RET) na 2680 CHF
- XLI, Ichthyosis, X-linked type (STS) 450 CHF
- PFIC3, Intrahepatic Cholestasis (ABCB4) na 2680-3030 CHF
- SMAX1, Kennedy (SBMA, AR) na 285 CHF
- KNO1, Knobloch, (COL18A1) na 1390 CHF
- Lactose intolerance (LCT) na 188 CHF
- LHON, Leber optical neuropathy 474 CHF
- LWD, Leri-Weill (SHOX) 450 CHF
- LFS, Li Fraumeni, (TP53) 1605-1955 CHF
- Marfan (FBN1) 3300 CHF
- NF1, Neurofibromatosis type I (NF1) na 3250 CHF
- PJS, Peutz-Jeghers (STK11) na 1605-1955 CHF
- PTEN Hamartoma Tumor syndrome (PHTS, Cowden, Hamartomas, BRRS, Proteus, PTEN) na 1605-1955 CHF
- PWS, Prader-Willi na 533 CHF
- Rendu-Osler-Weber (ROW) na @ 2900 CHF
- RETT syndrome (MECP2) 1310 CHF
- RSS, Russell-Silver syndrome (11p15) na 533 CHF
- Sick cell anemia (Drepanocytosis, HBB) 288 CHF
- UPD, Uniparental Disomy, Chr : \_\_\_\_\_ 470 CHF
- WVF, all types, na @ 960-2900 CHF
- WAGR, Wilms tumor (WT1) na 533 CHF
- Alpha-1-antitrypsin deficiency (A1AT)**
  - Genotyping P1\*SZ 210 CHF
  - Full sequencing of SERPINA1 na 645 CHF
- Ashkenazi mutations (rare disease carrier)**
  - Full screening (960 CHF) or
  - CFTR na Fragile-X na
  - Tay-Sachs+ FD+Fanconi+Canavan
  - von Gierke+Bloom+Niemann-Pick+ML-IV

Individual prices available upon request
- Ataxias**
  - Full screening (1765 CHF) or 185 CHF per gene +100 CHF
  - SCA1 na  SCA2 na  SCA3 na
  - SCA6 na  SCA7 na  SCA17 na
  - Friedreich  DRPLA na  FXTAS na
- Cardiac Arrhythmias (Channelopathies, CCP)**
  - SCN5A gene (Brugada) na 2680 CHF
  - KCNQ1 gene (QT-long) na 2680 CHF
  - KCNH2 gene (QT-long) na 2680 CHF
  - KCNE1, KCNE2, KCNJ2 genes na 1605 CHF

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 450 CHF
  - PMP22 gene sequencing (CMT1A) 960 CHF
  - MPZ gene sequencing (CMT1B) 1390 CHF
  - GJB1 gene sequencing (CMTX) 745 CHF

**Chromosomal Microdeletions**

- 22q11, MLPA 450 CHF
- Screening, recurrent microdeletions, MLPA 450 CHF
- Cystic Fibrosis (CF, CFTR)**
- CFTR: please indicate the ethnic origins of the patient at page 1
  - Screening frequent mutations (+IVS8 5T, CFTR-related disorders) 470 CHF
  - Full CFTR analysis (sequencing+ del/dup by MLPA) na 1820-2170 CHF
  - Hyperechogenic fetal bowel (frequent mutations CFTR in parents) 470 CHF
  - Hyperechogenic fetal bowel (frequent mutations CFTR +del/dup by MLPA in parents) 820 CHF
- Deafness**
  - DFNB1, congenital (locus DFNB1) na 665 CHF
  - Mitochondrial mutations na 645 CHF
- Endocrine Neoplasias, Pheochromocytoma, Paraganglioma (MEN, PCC, PGL)**
  - MEN1, Multi. Endoc. Neopl. type I (MEN1) 1605-1955 CHF
  - MEN2, Multi. Endoc. Neopl. type II (RET) 1605 CHF
  - PGL/PCC, Paraganglioma/Pheochromocytoma: Full sequencing (+MLPA) 2900 CHF (3950 CHF) or
    - SDHB gene  RET gene
    - SDHC gene na  SDHD gene

Individual prices available upon request

  - Von Hippel Lindau (VHL) 745-1095 CHF
- Familial Pneumothorax (Birt-Hogg-Dubé, BHD)** na
  - Frequent mutation, FLCN, exon 11 315 CHF
  - FLCN full gene analysis 2680-2930 CHF
- Familial Adenomatous Polyposis (FAP)**
  - Full Screening APC + MUTYH (2900-3600 CHF) or
  - APC gene 2895-3245 CHF
  - MAP (MUTYH, ex. 7,13) na 530 CHF
- FGFR3 (syndromes linked to)**
  - Achondroplasia 315 CHF
  - Craniosynostosis or Muenke 315 CHF
  - Hypochondroplasia 745 CHF
  - TD types I, II na 960 CHF
  - SADDAN na 315 CHF
- Fibrinopathies**
  - Afibrinogenemia (FGA, FGB, FGG) 1710 CHF
  - Dysfibrinogenemia (FGA, FGG) 530 - 1710 CHF
  - Hypofibrinogenemia (FGA, FGG) 530 - 1710 CHF
- Fragile X (FRAXA, FMR1)**
  - Diagnostic 285 CHF
  - Carrier testing 285 CHF
  - Premature ovarian failure (POI) 285 CHF
- Genetic sex**
  - Genetic sex determination 210 CHF
  - SRY search in a Turner 285 CHF
  - SRY sequencing 530 CHF
- Hemophilias**
  - HA, inversions F8 (IVS22, IVS1) 553CHF
  - HA, F8, complete analysis na 1605-1955 CHF
  - HB, F9, complete analysis 1605-1955 CHF
- Huntington disease (HD, HTT)** 285 CHF
  - Diagnostic
  - Presymptomatic (2 tubes please)
- Hereditary Periodic Fevers (HRF)**
  - Full Sequencing (8 genes) 2900 CHF
  - Full Screening Frequent Mutations (4 genes) 1820 CHF
  - FMF, MEFV gene 530 CHF
  - FMF, MEFV gene (complete sequencing) 1605 CHF
  - CAPS, NLRP3 gene 530 CHF
  - HIDS, MVK gene 530 CHF
  - TRAPS, TNFRSF1A gene 530 CHF

Whole Exome Sequencing and Targeted Gene Panel Analysis: see next page (page 4)
- HNPP (tomaculous neuropathy)**
  - Deletion CMT1A 450 CHF
  - PMP22 gene sequencing 960 CHF

**Lynch syndrome (HNPCC)**

- Full screening by NGS (2900-4165 CHF) or
- MLH1+PMS2 genes 2900-3600 CHF
- MSH2+MSH6 genes 2900-3600
- MSI (on tumor biopsy) 185 CHF
- BRAF1 V600E (on tumor biopsy) 315 CHF na

**Male infertility**

- CFTR+5T (frequent mutations) 470 CHF
- Y chromosome microdeletions (DAZ) 415 CHF

**Mitochondriopathies (ADNmt)**

- MELAS, MERRF, NARP Screening 472-660 CHF
- Deletions/Dup (muscle biopsy only) na 660 CHF
- Chromosome Mit sequencing na 1605 CHF

**Monogenic Diabetes (MODY, NDM)** na

- Targeted Exome Sequencing (gene panel) 2900-3300 CHF
- HNF4A gene (MODY 1) 1605-1955 CHF
- GCK gene (MODY 2) 1605-1955 CHF
- HNF1A gene (MODY 3) 1605-1955 CHF
- PDX1 gene (MODY 4) 530-880 CHF
- HNF1B gene (MODY 5) 1605-1955 CHF
- INS gene (MODY 10) 530-880 CHF
- KCNJ11 gene 530-880 CHF

**Neurological and Neuromuscular**

- SMA, Spinal Musc. Amyotrophy (SMN1) 450 CHF
- CADASIL (NOTCH3) na 1605 CHF
- DOPA-responsive dystonia (GCH1) na 1740 CHF
- Dravet syndrome (SCN1A) na 1605-1955 CHF
- EPM1, Unverricht-Lundborg (CSTB) na
- Southern+sequencing 1108 CHF
- FSHD, Facio-Scapulo-Humeral Dystrophy (only from EDTA blood), 660 CHF
- GLUT1 (SLC2A1) na 1955 CHF
- SPAST, Hered. Spastic Paraparesis (SPG4) 450 CHF
- STARTLE (Hyperekplexia, GLRA1) na 1605 CHF
- DMD, BMD, Muscular Dystroph Duchenne/Becker (DMD), deletions 800 CHF
- DM1, Myotonic Dystrophy of Steinert (DMPK) 285-875 CHF
- OPMD, Oculopharyngeal Muscular Dystrophy (PABPN1) na 315 CHF
- PD, Parkinson (PARK1 & 2, LRRK2) na 800 CHF
- TOR1A, Torsion Dystonia (DYT1) na 315 CHF

**Pancreatitis**

- CFTR+ IVS8 5T (frequent mutations) 470 CHF
- SPINK frequent mutation 215 CHF
- PRSS1 frequent mutations na 530 CHF

**Primary Ciliary Dyskinesia (PCD)** na

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

**Waardenburg (WS)** na

- Types I and III (PAX3 gene) 1605-1955 CHF
- Type II (MITF gene) na 1605-1955 CHF

**Miscellaneous (cf. additional informations)**

- DNA Extraction + Banking 61 CHF
- Circulating cell-free DNA Extraction na 61 CHF (only in Streck BCT or PAXgene blood DNA tubes)
- Out-of-list (OFAS) gene na x per exon 215 CHF, ax 2900-3250 CHF
- Specific/ Known familial mutation na 315 CHF
- Exclusion of maternal contamination in fetal sample (amnio-, choriocentesis) na 185 CHF
- Transfer of DNA to an external laboratory (please provide specifics below).

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

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, costs...). All prices are in Swiss Franc (CHF), not including DNA extraction cost (see DNA banking) nor possible variant verification by Sanger sequencing. For prenatal tests (urgent), an additional charge applies (200 CHF). 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 61 CHF

### HIGH THROUGHPUT SEQUENCING FOLLOWED BY BIOINFORMATIC ANALYSIS OF 1 TO 10 GENES (2900 CHF) @

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).

Mitochondriopathies

Cardiac Channelopathies (Arrhythmias, CCP) \*

Cardiomyopathies \*

Primary Ciliary Dyskinesia (PCD) \*

FGF receptor-associated dysplasias

Duchenne et Becker dystrophinopathies

Hereditary periodic fevers \*

Wilson's disease

Hereditary neoplasia

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 (3300 CHF – 3800 CHF) @

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 3300 CHF

> 100 genes 3800 CHF

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

11-100 genes 3300 CHF

**Cardiomyopathies \*na**

11-100 genes 3300 CHF

**Primary Ciliary Dyskinesia (PCD) \***

11-100 genes 3300 CHF

**Ehlers-Danlos**

11-100 genes 3300 CHF

**Epilepsy \***

11-100 genes 3300 CHF

> 100 genes 3800 CHF

**Diseases related to coagulation, blood and immune system disorders**

11-100 genes 3300 CHF

> 100 genes 3800 CHF

**Muscular dystrophies due to protein disorders associées à la dystrophine**

11-100 gènes 3300 CHF

**Neuromuscular et neurodegenerative diseases**

11-100 genes 3300 CHF

> 100 genes 3800 CHF

**Diseases related to skin, connective tissue or bones**

11-100 genes 3300 CHF

> 100 genes 3800 CHF

**Metabolic and endocrine diseases**

11-100 genes 3300 CHF

> 100 genes 3800 CHF

**Ophthalmologic diseases**

11-100 genes 3300 CHF

### ADDITIONAL ANALYSES @

Additional bioinformatic analysis  1-10 genes: 600 CHF  11-100 genes : 1000 CHF  more than 100 genes : 1500 CHF

Other additional analyses  Sanger sequencing 215 CHF  MLPA 350 CHF  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: \_\_\_\_\_