

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



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° :**

DIAGMOL

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

Head of Genetic Medicine Division : Prof. Marc ABRAMOWICZ
Laboratory managers (medical genetics FAMH):
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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

PHYSICIAN

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

[Empty box for Physician information]

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

[Empty box for Copy to other physician information]

« 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:

[Empty box for index case name]

CLINICAL INFORMATIONs given by the physician:

[Empty box for Clinical information]

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)

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.sgmq.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.
- cl Depending on the choice of technique, the analysis may or may not be accredited.

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) * 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) * 631 CHF
- FG (Keller syndrome, MED12) * na 530 CHF
- HBLRG, Gilbert syndrome (UGT1A1) * 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) * cl 2680-3030 CHF
- SMAX1, Kennedy (SBMA, AR) na 285 CHF
- KNO1, Knobloch, (COL18A1) na 1390 CHF
- Lactose intolerance (LCT) * 188 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
- Non-invasive prenatal diagnostic of monogenic diseases (contact us in advance) * 615-2900 CHF
- PJS, Peutz-Jeghers (STK11) * na 1605-1955 CHF
- PTEN Hamartoma Tumor syndrome (PHTS, Cowden, Hamartomas, BRRS, Proteus, PTEN) * 1605-1955 CHF
- PWS, Prader-Willi na 533 CHF
- Rendu-Osler-Weber (ROW) * cl @ 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
- VWF, all types, * na @ 960-2900 CHF
- WAGR, Wilms tumor (WT1) * na 533 CHF

Alpha-1-antitrypsin deficiency (A1AT)

- Genotyping PI*S/Z 286 CHF
- Full sequencing of SERPINA1 na 645 CHF

Ashkenazi mutations (rare disease carrier)

- Full screening * (865 CHF) or
 - CFTFR Fragile-X
 - 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) cl 2680 CHF
 - KCNQ1 gene (QT-long) cl 2680 CHF
 - KCNH2 gene (QT-long) cl 2680 CHF
 - KCNE1, KCNE2, KCNJ2 genes cl 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 (with 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) * 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 cl 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) na

- 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) * 553 CHF
- HA, F8, complete analysis cl 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 see next page (page 4) or
- MLH1+PMS2 genes 2900-3600 CHF
- MSH2+MSH6 genes 2900-3600
- MSI (on tumor biopsy) 315 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)

- Leber optic neuropathy (LHON) 530 CHF
- Cytopathy MELAS, MERRF, NARP 472-660 CHF
- Deletions/Dup (muscle biopsy only) * 660 CHF
- Chromosome Mit sequencing 1605 CHF

Monogenic Diabetes (MODY, NDM) *

- Whole Exome Sequencing and Targeted Gene Panel Analysis: see next page (page 4) or
- HNF4A gene (MODY 1) cl 1605-1955 CHF
- GCK gene (MODY 2) cl 1605-1955 CHF
- HNF1A gene (MODY 3) cl 1605-1955 CHF
- PDX1 gene (MODY 4) cl 530-880 CHF
- HNF1B gene (MODY 5) cl 1605-1955 CHF
- INS gene (MODY 10) cl 530-880 CHF
- KCNJ11 gene cl 530-880 CHF

Neurological and Neuromuscular

- SMA, Spinal Musc. Amyotrophy (SMN1) 450 CHF
- CADASIL (NOTCH3) * 1605 CHF
- DOPA-responsive dystonia (GCH1) * na 1740 CHF
- Dravet syndrome (SCN1A) * na 1605-1955 CHF
- EPM1, Unverricht-Lundborg (CSTB) *
- Southern+sequencing 1108 CHF
- FSHD1, 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
- TOR1A, Torsion Dystonia (DYT1) * na 315 CHF

Pancreatitis

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

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) 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 * per exon 215 CHF, ax 2900-3250 CHF
- Specific/ Known familial mutation * 315 CHF
- Exclusion of maternal contamination in fetal sample (amnio-, choriocentesis) * 185 CHF
- Transfer of DNA to an external laboratory (please provide specifics below) and complete the ECA forms for internal requests.

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, 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).

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 (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

Diabetes, monogenic (MODY, NDM) *

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

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

> 100 genes 3800 CHF

Diseases related to urogenital system, fertility / sterility

11-100 genes 3300 CHF

> 100 genes 3800 CHF

Hereditary neoplasia

11-100 genes 3300 CHF

> 100 genes 3800 CHF

Sensorimotor neuropathies

(CMT, HNPP, amyloid polyneuropathy)

11-100 genes 3300 CHF

Kallman syndrome

11-100 genes 3300 CHF

Marfan syndrome and other thoracic aorta syndromes

11-100 genes 3300 CHF

Mendelian syndrome with growth disorder

11-100 genes 3300 CHF

> 100 genes 3800 CHF

Developmental disorders *

11-100 genes 3300 CHF

> 100 genes 3800 CHF

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

11-100 genes 3300 CHF

> 100 genes 3800 CHF

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 3300 CHF

> 100 genes 3800 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: