

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.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)

[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 BCI 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, Icthyosis, X-linked type (*STS*) 450 CHF
 PFIC3, Intrahepatic Cholestasis (*ABC4*) * 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
 Sickle 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* 645 CHF

Ashkenazi mutations (rare disease carrier)

- Full screening * (865 CHF) or
 CFTR 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-short) 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 *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
 Thanatophoric dysplasia, types I, II 960 CHF
 SADDAN 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 CHF
 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
 C9orf72 (ALS, FTD) * 470 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 BCI 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)

Transfer of DATA of NGS to an external laboratory (please provide specifics below) and complete the DATA exchange form.

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: _____