REQUEST FORM: CONSTITUTIONAL GENETIC ANALYSIS

LNS LUXEMBOURG
LABORATOIRE NATIONAL DE SANTE
NATIONAL CENTER OF GENETICS
MOLECULAR DIAGNOSTICS

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Fax. (+352) 28 100 - 542
ncg-avis-prescription-genetique@lns.etat.lu
Formulaire disponible sous www.lns.lu

SAMPLE COLLECTION INFORMATION *(or provide transfer sheet)*

- 2x EDTA (5ml)
- 1x Heparine (5ml) (If Karyotype)

-Transport at Room T°

Date/time of sampling:

On ..... **at:** .....

Sent on: .....

Material :

☐ **Peripheral blood**, number of tubes: .....

☐ **Other samples**, precise : .....

LABEL / LNS BARCODE

ETIQUETTE LNS

PRESCRIBER

Prescriber Last name – First name

Address – Country

Phone / Direct phone | Fax

Prescription date | Signature / Stamp

Copy (name and address)

Results will **only** be sent to the prescriber (and doctor in copy if requested)

PATIENT

Birth name | First name

Marital name | Sex

Date of birth | Matricule

Address - Country

Patient affiliated to CNS/UCM ☐ Yes ☐ No*

*If not-affiliated to CNS/UCM, the patient will receive an invoice from the laboratory which they can send to their insurance company.

INFORMATION

This form summarizes the most common indications and genetic tests classified by clinical area or speciality.

Please complete the request form and the informed consent below. As a reminder, the signatures of the patient AND the prescriber are mandatory for genetic testing.

For any question regarding a genetic analysis that you wish to prescribe, particularly those not listed in this form, you can reach us at the following address: **ncg-avis-prescription-genetique@lns.etat.lu** .

Note that specific request form or genetic consultation **may be required** for some genetic tests.

Clinical data are mandatory. Please **specify the indication** justifying the request. This will allow the laboratory team, after consultation with the referring geneticist, **to better target the appropriate genetic analysis for the patient**.

For **a request for constitutional oncogenetic** test or **prenatal testing please use the appropriate form** available on our website.

Clinical data

Urgent Analysis : ☐ **Ongoing pregnancy** ☐ **Neonate** ☐ **Other reason, specify :** .....

☐ Index case

☐ Related - Name of index case: ..... Link to index case: ☐Father ☐Mother ☐Brother/Sister ☐Other, specify:.....

Symptomatic patient : ☐ Yes ☐No*

☐ Known mutations : attach report of the index case

☐ **DNA extraction and storage**

**The prescription of genetic tests for pre/asymptomatic patient is limited to geneticist after genetic counselling*

INFORMED CONSENT

By signing below, I consent to the genetic analysis as indicated on this request form to determine the genetic cause of the medical condition mentioned. I agree to send the results to the prescribing physician.

I was informed by the signing doctor of the medical necessity, potential benefits and limitations of the planned genetic test. In addition, the possible consequences of communicating the test result (e.g., psychological burden) have been discussed.

With your consent, unused sample material and tests results will be stored. - I accept that they may be used to verify the results obtained, for subsequent analyses and as part of the laboratory's quality assurance. - I accept that they may be used for research and scientific publication in an anonymous form or as part of university teaching.	☐ Yes ☐ No ☐ Yes ☐ No

I am aware that my consent applies to me and/or my minor child(ren), and that I may withdraw this consent at any time without having to provide reasons.

SPACE INTENDED FOR LABORATORY

TAMPON ACCEPTABILITE LNS

Patient signature

Date / Time : .....

Signature : .....

Prescripteur signature

Date / Time : .....

Signature : .....

The LNS processes the data collected to carry out analyses and transmit the results. To learn more and to exercise your rights, refer to the LNS personal data protection policy: https://lns.lu/donnees-personnelles/

[MOLGEN-PR1-FOR-02073] [ENG] (A) ; v2

Patient name:

HLA GENOTYPING, PHARMACOGENOMICS AND SUSCEPTIBILITY POLYMORPHISMS

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Celiac disease susceptibility	☐ HLA-DQ2/DQ8
☐ Meulengracht disease (Gilbert syndrome)	☐ UGT1A1
☐ Behçet disease	☐ HLA-B51
☐ Uveitis	☐ HLA-B27
☐ Spondyloarthritis	☐ HLA pre-transplantation – Please indicate the laboratory and attach its specific request form
☐ Pre-transplantation HLA-typing	
☐ Other:	☐ other: .....

REPRODUCTIVE DISORDERS and PRECONCEPTIONAL SCREENING

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Male Infertility ☐ Secretory Azoospermia ☐ Obstructive azoospermia ☐ OATS ☐ Cryptozoospermia	☐ Karyotype (*please send us an heparin tube*)
☐ Female infertility ☐ Primary ovarian Insufficiency (POI) ☐ Other: Specify.....	☐ Chromosome Y microdeletion (AZF) ☐ CGG repeat expansion FMR1 gene (Fragile-X) (premutation FMR1). ☐ Most frequent mutations of CFTR. ☐ CYP21A2 sequencing (congenital adrenal hyperplasia).
☐ Recurrent pregnancy loss(>2) Name and/or matricule of the partner:.....	☐ Other: .....
☐ pre-IVF or pre-PGD screening Name and/or matricule of the partner:.....	
☐ Other:	

HEMATOLOGY

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Systematic screening (genetic counseling in the context of pregnancy, ethnicity) : ☐ ongoing pregnancy / ☐ MAP screening	☐ hemoglobin analysis (Hb electrophoresis, HbA2 and HbF dosage, molecular analysis of α- et β-thalassemia) (recent blood count and ferritin value mandatory)
☐ Family history (specify):	☐ Factor V-Leiden (p.R506Q)
☐ microcytosis and/or unexplained polycythemia	☐ Factor II - Prothrombin (G20210A)
☐ Follow up of known hemoglobinopathy (specify):	☐ PAI-1 (4G/5G gene promoter)
☐ Incidental identification of an Hb variant by HbA1C testing	☐ MTHFR (c.C677T and c.A1298C)(*If homocysteine plasma level > 50 µmol/L*)
☐ Etiological assessment of hemolytic anemia	☐ Other: .....
☐ Thrombophilia	
☐ Other:	

PEDIATRICS and NEUROPEDIATRICS

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Autism spectrum disorder	☐ Fragile X syndrome (CGG repeat expansion of the FMR1 gene).
☐ Developmental delay/ intellectual disability (if IQ < 70)	☐ array CGH (*please add peripheral blood (EDTA) from both parents*).
☐ Epilepsy	☐ Study of methylation on SNRPN locus (Prader-wili/Angelmann).
☐ Neonatal hypotonia	☐ Epilepsy panel.
☐ Myopathy and muscular weakness.	☐ Myopathy panel.
☐ Steinert disease	☐ DMD gene analysis. ➢ Search for deletions/duplications. ➢ *Complete gene sequencing*
☐ Duchenne/Becker disease (provide result of the CPK blood test)	☐ Copy number of SMN1 and SMN2 genes.
☐ Spinal muscular atrophy (SMA)	☐ Repeat expansion of DMPK and/or PROMM gene (Myotonic dystrophy).
☐ Cystic fibrosis (attach sweat test if available)	☐ CFTR ➢ *Most frequent mutations* ➢ *Complete gene sequencing (after discussion with the laboratory)*
☐ Growth delay / short stature	☐ Other: .....
☐ Other:	

ADULT NEUROLOGY

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Dementia : age of onset.....	☐ Epilepsy panel.
☐ Parkinson Disease	☐ Myopathy panel.
☐ Epilepsy	☐ CGG repeat expansion FMR1 gene.
☐ Ataxia /Chorea/ Dyskinesia/Dystonia : Age of onset (please specify).....	☐ Movement disorders panel. ☐ ALS panel.
☐ Myopathy. Age of onset (please specify).....	☐ repeat expansion C9orf72 (sampling Monday/Tuesday, shipping < 24h)
☐ Huntington disease.	☐ Analysis of PMP22 locus.
☐ Peripheral Neuropathy Age of onset : (specify)..... ☐ Sensitive ; ☐ motor ; Symmetrical ?: Yes ☐ No ☐	☐ Neuropathy panel ☐ NOTCH3 gene. ☐ HLA-DQB1*06:02
☐ Amyotrophic lateral sclerosis (ALS)	
☐ Narcolepsy	☐ Other: .....
☐ Hemiplegic Migraine	
☐ CADASIL	
☐ Other:	

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Patient name:

CARDIOLOGY

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Dilated / hypertrophic/ arrhythmogenic cardiopathy	☐ GLA gene (Fabry)
☐ Fabry disease	☐ TTR gene (Amyloïdosis)
☐ Cardiac amyloïdosis	☐ Arrhythmia panel.
☐ Long QT syndrome/ Brugada syndrome	☐ Cardiomyopathy panel.
☐ Aneurysm/ aortic dissection	☐ Connective tissue panel (*Marfan syndrome,Loeys-Dietz syndrome,…*)
☐ Other: ……	☐ Other: ……

GASTROENTEROLOGY / HEPATHOLOGY

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Hereditary chronic pancreatitis	☐ *CFTR:*
☐ Alpha1 Anti-trypsin deficiency	➢ *Most frequent mutations*
☐ Cholestasis / chronic diarrhea	➢ *Complete gene sequencing (after discussion with the laboratory)*
☐ Hemochromatosis / Iron overload	☐ Screening of SERPINA1 gene
☐ Wilson disease	☐ HFE : most frequent mutations (C282Y, H63D)
☐ Other: ……	☐ ATP7B gene (Wilson disease)
	☐ Chronic pancreatitis panel.
	☐ Other: ……

ENDOCRINOLOGY and METABOLIC DISEASES

CLINICAL INDICATION	REQUESTED TEST(S)
☐ MODY Diabetes	☐ MODY panel
☐ Familial hypercholesterolemia	☐ Familial hypercholesterolemia panel
☐ Congenital adrenal hyperplasia, 21-hydroxylase block	☐ Disorder of sexual development panel
☐ Hypogonadotropic hypogonadism	☐ Hypogonadism panel
☐ Hypergonadotropic hypogonadism (primary ovarian insufficiency)	☐ Short stature panel.
☐ Disorder of sexual development (attach karyotype result)	☐ Analysis of *SHOX gene (MLPA/sequencing)*
☐ Short stature (attach karyotype result)	☐ Analysis *SRY gene*
☐ Other: ……	☐ Other: ……

NEPHROLOGY

CLINICAL INDICATION	REQUESTED TEST(S)
☐ Polycystic kidney disease.	☐ Polycystic kidney panel.
☐ Alport syndrome.	☐ Lithiasis/nephrocalcinosis panel
☐ Lithiasis/nephrocalcinosis.	☐ Tubulopathy panel
☐ Hemolytic Uremic Syndrome – HUS.	☐ Alport panel
☐ Focal segmental glomerulosclerosis-FSGS.	☐ FSGS panel.
☐ Nephronophtisis.	☐ *PKD1/2 panel*
☐ Interstitial tubulopathy.	☐ *CD46 gene (HUS)*
☐ Familial hyperaldosteronism.	
☐ Other: ……	☐ Other: ……

OTHER INDICATIONS

Please specify the indication or the clinical context	**Please specify the requested test(s) :** *Gene / Gene panel / Specific Mutation / Other*
……	……
……	……
……	……