Institute of Genomic Medicine and Rare Disorders
Semmelweis University
H-1083 Budapest, Tömő u. 25-29. Tel:+36-1-4591492
Email: molneur@med.semmelweis-univ.hu,
molneur@gmail.com
http://semmelweis.hu/genomikai-medicina/

GENETIC TEST REQUEST FORM

Name:		Maiden name:
Social security number:		Mother's name:
Date of birth:		
Address:		
Requesting physician's name:		Requesting physician's stamp number:
Requesting physician's email:		
OEP code:		Diagnosis (BNO code):
Sample type:	◊ EDTA blood (9 ml) ◊ nerve/muscle tissue:	◊ other:
Date of sampling:		Ambulatory Registry Number:

Please enclose the ambulatory sheet/medical report containing the patient's clinical data.

DECLARATION OF CONSENT

Planned intervention: ……..

Name of patient:
Name of legal representative/relative:

Health condition, medical opinion:
……..
……..

Possible advantages and risks of completing / failing the recommended medical examinations (side effects, complications):
……..
……..

Possible advantages and risks of completing / failing the recommended interventions (side effects, complications):
……..
……..

Surgical and non-surgical possibilities:
……..

Medical examination and expectable outcome of the intervention, and the probability of the success of that:
Asking for information on complications with a probability of under 1% **YES NO**
……..
……..

Planned date and time of the medical examinations and interventions:…..……...
(The patient is aware of the possibility of variation. In case of that, he or she gets informed.)

Expectable fee of the care:……..

Questions of the patient/legal representative and answers to them:
……
……
…….

I have been informed about the risks and possible frequent complications and expectable consequences of the recommended intervention, I have received personalized answers to my verbal/written questions, and I had time enough for making a defined decision about the way of the treatment.

I allow to complete necessary intervention other than the planned one occurring during the operation, and to expand the operation in case the medical specialist considers that necessary or an urgent necessity requires that.

I accept that unpredictable complication can happen also in case of a professional treatment, which has negative effect on the expectable results and recovery time.

I accept that expect for the life support and rescue operation defined by the physician, I have the right to reject any of the recommended care. In this case, I make a written declaration about the fact of the rejection. At the request of legal representative, in case of an incapable patient or a patient with limited incapacity, care with an expectable consequence of a serious or irrevocable damage in the patient's health condition is not allowed to reject.

I Agree, Disagree (please underline) with the planned intervention to be completed on me.

I Agree, Disagree (please underline) with preserving any cell parts, tissues, organs removed for diagnostic and health care purposes related to the medical examination ad intervention by the Semmelweis University, and by the use of them, to execute scientific and medical research.

I allow my physician and other patient care service to handle my personal health care data in order to contribute the effective patient care. My physician has informed me that Semmelweis University maintains an integrated informatics health care data handling system, so information share can happen in the interest of my own care.

Budapest, 20…. ……..

Patient's or legal representative's signature	Date and witness signature

Institute of Genomic Medicine and Rare Disorders
Semmelweis University
H-1083 Budapest, Tömő u. 25-29. Tel:+36-1-4591492
Email: molneur@med.semmelweis-univ.hu,
molneur@gmail.com
http://semmelweis.hu/genomikai-medicina/

Diseases of Central Nervous System

Monogénes stroke syndroma
- ◊ CADASIL (NOTCH3) gene mutation screening
- ◊ MELAS (m.3243A>G) point mutation analysis

Stroke risk factor test
- ◊ Leiden Factor V mutation screening
- ◊ ApoE genotyping
- ◊ MTHFR (methionine tetrahydrofolate reductase) mutation screening (c.677 C>T)
- ◊ Thrombocyte glycoprotein receptor polymorphism exam: GPIb alpha-VNTR

Trinucleotide repeat disorders
- ◊ Huntington's disease (differential diagnosis)
- ◊ Fragile X syndromes
  - ◊ Classic Fragile X syndrome
  - ◊ FXTAS syndrome
  - ◊ POI/POF syndrome
- ◊ Spinocerebellar Ataxia profile (ataxia differential diagnostic) or
  - ◊ SCA1 (ATXN1) gene
  - ◊ SCA2 (ATXN2) gene
  - ◊ SCA3 (ATXN3) gene
  - ◊ SCA6 (CACNA1) gene
  - ◊ SCA7 (ATXN7) gene
- ◊ Dystrophy myotonic type 1

Parkinson disease (familiar or early onset)
- ◊ PARK2 gene analysis
- ◊ LRRK2 gene analysis
- ◊ PINK1 gene analysis
- ◊ DJ1 gene analysis

Dystonia
- ◊ DYT1 (Torsin A) gene deletion screening

Iron accumulation in basal ganglia
- ◊ Coeruloplasmin e(CP) gene mutation analysis

Neurodegeneration with brain iron accumulation
- ◊ BPAN gene analysis
- ◊ COASY gene analysis
- ◊ MPAN (C19orf12) gene analysis
- ◊ PANK2 gene analysis
- ◊ PLA2G6 gene analysis

Hereditary Alzheimer disease
- ◊ APP gene analysis
- ◊ Presenilin-1 (PS1) gene analysis
- ◊ Presenilin-2 (PS2) gene analysis

Frontotemporal dementia
- ◊ TAU protein (MAPT) gene analysis
- ◊ Granulin (GRN) gene analysis

Prion diseases
- ◊ PRNP3 gene analysis

Mental decline, dementia, atherosclerosis risk factor
- ◊ ApoE genotyping
- ◊ TREM2 gene analysis

Migraine risk factor test
- ◊ MELAS (m.3243A>G) point mutation analysis
- ◊ MTHFR mutation analysis (c.677 C>T)
- ◊ CADASIL (NOTCH3) gene point mutation screening

Non syndrome hearing loss
- ◊ Connexine 26 (GJB2) gene analysis

Optic atrophy
- ◊ OPA1 gene analysis
- ◊ Leber Hereditary Optic Neuropathy (mtDNS)

Beta oxidation disorder
◊ MCAD gene (m.985 A>G) genotyping

Depression, posttraumatic stress
- ◊ 5HTT receptor gene genotyping

Mitochondrial DNA disorders
- ◊ PEO - mtDNS deletion screening
- ◊ Myopathy - mtDNA deletion screening
- ◊ mtDNA depletion syndrome (only from muscle, up to the age of 4)
- ◊ MELAS (m.3243A>G) mutation analysis
- ◊ NARP (m.8993 T>C, T>G) mutation analysis
- ◊ MERRF (m.8344 A>G,) mutation analysis
- ◊ LHON (m.4360 G>A, m.11778 G>A, m.14484 T>C) point mutation analysis
- ◊ Whole mtDNA sequencing (from muscle tissue with special agreement)

Intergenomic communication disorders (can be requested only in case of mtDNA deletion/depletion in the muscle tissue)
- ◊ mtDNA depletion syndrome (only from muscle, up to the age of 4)
- ◊ Multiplex mtDNA deletion screening
- ◊ Alpers syndrome (POLG1 gene analysis)
- ◊ SANDO (POLG1 gene analysis)
- ◊ PEO syndrome (POLG1 gene analysis)
- ◊ PEO syndrome (Twinkle gene analysis)
- ◊ PEO syndrome (ANT1 gene analysis)
- ◊ PEO syndrome (RRM2B gene analysis)
- ◊ Depletion syndrome (TK2 gene analysis)
- ◊ SMA like image (TK2 gene analysis)
- ◊ Myopathy (TK2 gene analysis)

Hypertrophic cardiomyopathy with COX negative fibres (infancy)
- ◊ SCO2 gene analysis

Neuromuscular Disorders

Myoglobinuria
- ◊ CPT-II (carnitin-palmitoyl-transferase II) mutation analysis (c.C338T, c.C149A hot spot)

Congenital Myasthenia Syndrome

◊ CHNRE Romani founder gene mutation analysis

Dystrophy myotonic type 1

◊ DMPK gene analysis

Facioscapulohumeral (FSHD) muscular dystrophy

◊ FSHD1A gene deletion analysis

Duchene-Becker muscular dystrophy

◊ Dystrophin gene analysis

Limb girdle (LGMD) muscular dystrophy DNA diagnostic

◊ LGMD2A (CAPN3 - 550 del A)
◊ LGMD2c (γ SG - p. C283Y)
◊ LGMD2I (FKRP c.826 C>A)

Muscular dystrophy protein diagnostic (from muscle)

◊ Dystrophin Western blot
◊ LGMD autosomal dominant form: (caveolin, myotilin) Western blot
◊ Autosomal recessive form: (dysferlin, calpain, sarcoglycans) Western blot

Spinal muscular dystrophy

◊ SMN1 gene deletion analysis

Spinal muscular dystrophy 1

◊ SCO2 gene analysis

Hereditary neuropathies

◊ Congenital cataracta facial dysmorphy neuropathy (CCFDN) founder mutation
◊ Lom (NDRG1) neuropathy founder mutation
◊ Hereditary neuropathy pressure palsy (HNPP) PMP22 deletion test
◊ Multiplex tunnel syndrome: PMP22 deletion
◊ Charcot Marie Tooth-I. (demyelinisatio type of neuropathy) profile: PMP22 duplication, connexin 32, MPZ, EGR2 gene analysis
◊ Charcot Marie Tooth-II (axonal type of neuropathy) profile: mitofusin, MPZ, Connexin 32 gene analysis
◊ Dejerine Sottas neuropathy MPZ, EGR2 gene analysis
◊ Congenital hypomyelination syndrome MPZ, EGR2, Connexin 32 gene analysis
◊ Autosomal dominant hereditary neuropathies: (PMP22 deletion/ duplication, MPZ, EGR2 gene analysis)
◊ X chromosomal hereditary neuropathy - Connexin 32 (GJB1) gene analysis

Pharmacogenomic Exams

Aminoglycoside-induced deafness

◊ (m.1555 A>G) mutation analysis

Azatioprine toxicity

◊ MTHFR mutation analysis

Citalopram side effect

◊ CYP2C19*2 genotyping

Clopidogrel effectiveness

◊ CYP2C19*2 genotyping

Diazepam side effect

◊ CYP2C19*2 genotyping

Lanzoprazol side effect

◊ CYP2C19*2 genotyping

Omeprazole side effect

◊ CYP2C19*2 genotyping

Pantoprazole side effect

◊ CYP2C19*2 genotyping

Statin-induced myopathy

◊ SLCO1B1, KIF6, COQ2, ATP2B1 genotyping

Ticagleror side effect

◊ CYP2C19*2 genotyping

Valproate-induced hepatotoxicity

◊ POLG1 gene SNPs exam

Non-supported, self-financing exams

Molecular cytogenetic tests (aCGH)

◊ Multiplex congenital minor anomalies
◊ Delayed global development/mental retardation with or without dysmorphic features
◊ Autism spectrum disease

Exams by new-generation sequencing

Panel tests

◊ Hereditary spastic paraparesis (51 genes)
◊ ALS panel (50)
◊ Autism panel (103 gene)

Whole exome test

Information on the costs of non-supported exams can be requested at molneur@med.semmelweis-univ.hu.

Please fill in the request form correctly and add all the clinical data, otherwise the exams will not be performed until the missing information arrives.

Date:

Physician's signature **stamp**