Institute of Genomic Medicine and Rare Disorders
Semmelweis University

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GENETIC TEST REQUEST FORM (2025)

Name:	Maiden name:
Social security number:	Mothers name:
Date of birth:	Address:
Name of requesting physician:	Stamp number:
e-mail of requesting physician:	Request NEAK code:
Diagnosis (BNO code):	
Date of sample:	Ambulatory Registry Number:

Sample type: ◊ EDTA blood (9 ml) ◊ other: ◊ nerve/muscle tissue:

Please aid our work by filling out below! Thank you!

Family anamnesis: Age at first symptoms: years

HPO of main symptom: BNO of accompanying illnesses:
Short clinical summary:

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

Declaration of Consent for genetic examination and biobank storage.

I authorise the isolation and storage of DNA from my biological sample and to carry out molecular biology tests from the DNA sample to determine factors predisposing to my illness and for genetic variants causative to my illness.

……..
(please specify the illness)

I agree to blood/tissue sample taking for molecular biological diagnostic, research or quality improvement purposes. The examination allows for the diagnosis of the disease or the identification of genetic factors predisposing to the illness or the efficiency or side effect of certain medications

I understand, that the result of the examination will not provide sufficient information in case of the following reasons or their combination: 1) Missing blood/tissue sample of relevant family member 2) The biological markers on hand are not informative, 3) Technical reasons.
In the case of research generating relevant information with respect to my health state/future illnesses:

- □ ◊ I wish to be informed
- □ ◊ I do not wish to be informed

I understand that I'm informed of the results of the genetic examination personally in the form of genetic counselling.

My DNA samples and clinical data can be stored in the DNA Bank and Register of the Institute of Medicine and Rare Disorders under appropriate security and data protection protocols.

I agree, that my DNA sample (my child's DNA sample) can be used by SE Institute of Medicine and Rare Disorders or a different molecular biology laboratory for research or quality improvement purposes under appropriate data protection protocols. I understand that information regarding my person (my child) are treated confidentially. My personal data will be not provided to other Institutes by SE until I make a separate written statement of this.

I will receive no financial benefits if my sample is used for research or quality improvement purposes.

With my signature I declare that I understood the information above and I participate in the molecular genetic examination and Biobanking having accepted these. The DNA sample is stored free of charge in the Institute for a minimum of 5 years and will be provided to you at any time should you require it for other disease aspects. The consent does not exempt the examiners or the Institute concerned from legal and professional obligations. If you require further information with respect to the above, please do not hesitate to contact your attending physician or genetic counsellor.

Signature of patient (or legal representative)	Date and signature of witness

Diseases of the Central Nervous System

Hereditary arteriopathy
- ◊ Hot spot analysis
  - o NOTCH3 gene exon 3-4 (CADASIL)
- ◊ Arteriopathy panel (NGS)
  - o NOTCH3, CTSA, HTRA1, COL4A1, BSCL2 genes

Fragile X associated syndromes
- ◊ FXS/FXTAS/FXPOI - FMR1 gene repeat number determination

Huntington disease
- ◊ HTT gene repeat number determination

Kennedy syndrome/SBMA
- ◊ AR gene repeat number determination

Ataxias
- ◊ Spinocerebellar Ataxia gene repeat number det.
  - o SCA1 (ATXN1 gene)
  - o SCA2 (ATXN2 gene)
  - o SCA3 (ATXN3 gene)
  - o SCA6 (CACNA1 gene)
  - o SCA7 (ATXN7 gene)
  - o SCA17 (TRP gene)
- ◊ Friedreich ataxia - FXN gene repeat number det.
- ◊ Late Onset Ataxias repeat number det.
  - o SCA27B (FGF14 gene)
  - o CANVAS (RFC1 gene)
- ◊ Ataxia panel (NGS)
  - o ATM, SACS, SETX, ANO10, APTX, SYNE1, CACNA1A, COQ4, COQ8A genes
- ◊ POLG1 gene sequencing (SANDO)

Parkinson's disease (familiar or late onset)
- ◊ Copy number examination (MLPA)
  - o SNCA, PARK2, PINK1, PARK7, ATP13A2, LRRK2 genes
- ◊ Hereditary Parkinson's disease panel (NGS)
  - o SNCA, PARK2, PINK1, PARK7, ATP13A2, LRRK2, GBA1 genes

Dystonia
- ◊ DYT1 (Torsin A) gene deletion test

Neurodegeneration with brain iron accumulation (NBIA)

Copy number examination (MLPA)
- o PANK2, PLA2G6 genes
- o NBIA panel (NGS)
- o C19orf12 (MPAN), WDR45 (BPAN), PANK2, PLA2G6, CP, FTL, COASY, FA2H, CRAT, REPS1 genes

Monogenic Dementias/ Dementia Risk Factors
- ◊ C9ORF72 gene repeat number determination
- ◊ Frontotemporal dementia panel (NGS)
  - o GRN, TARDBP43, MAPT genes
- ◊ Monogenic Alzheimer panel (NGS)
  - o PS1, PS2, APP genes
- ◊ ApoE genotyping

Creutzfeldt-Jakob disease
- ◊ PRNP3 gene sequencing

Non syndromic hearing loss
- ◊ Connexin 26 (GJB2) gene sequencing

N. Optical atrophy
- ◊ OPA1 gene sequencing
- ◊ LHON (m.4360 G>A, m.11778 G>A, m.14484 T>C) analysis

Beta oxidation disorder (ACADMD)
- ◊ MCAD gene sequencing

Lysosomal disorders (Can only be requested in case of pathogenic enzyme activity/substrate tests)
- ◊ Fabry disease (GLA gene)
- ◊ Gaucher disease (GBA1 gene)
- ◊ Pompe disease (GAA gene)

Mitochondrial DNA disorders
- ◊ MELAS (m.3243A>G) analysis
- ◊ NARP (m.8993 T>C, T>G) analysis
- ◊ MERRF (m.8344 A>G) analysis
- ◊ LHON (m.4360 G>A, m.11778 G>A, m.14484 T>C) analysis
- ◊ mtDNA deletion screening (PEO, myopathy)
- ◊ mtDNA depletion syndrome (**muscle only**)
- ◊ Whole mtDNA sequencing (with LHON indication, following personal consultation, from blood/muscle.)

Nuclear mitochondrial genes
- ◊ Gene panel (NGS)
  - o POLG1, OPA1, TWNK, RRM2B, ANT1, SCO2, TK2 genes

Hereditary Spastic Paraparesis (HSP)
- ◊ Copy number examination (MLPA)
  - o ATL1 (SPG3A), SPAST (SPG4) genes
- ◊ HSP panel (NGS)
  - o SPAST (SPG4), SPG7, SPG11, AP4M1 genes

Intellectual Impairment/Autism Spectrum Disorder

- ◊ Copy number examination (MLPA)
  - o Microdeletion MLPA (P245)
  - o ASD Microdeletion MLPA (P343)
- ◊ RNU4-2 gene sequencing
- ◊ Fragilis X associated disorder: FXS

NEUROMUSCULAR DISEASES

Facioscapulohumeral muscle dystrophy (FSHD)

- ◊ FSHD1A gene deletion analysis
- ◊ FSHD2 methylation/haplotype analysis

Duchenne-Becker type muscle dystrophy

- ◊ DMD gene deletion/duplication (MLPA)
- ◊ Deletion/duplication segregation: Exon(s): ☐
- ◊ DMD gene sequencing

Limb Girdle Muscle Dystrophy (LGMD)

- ◊ Hot spot analysis:
  - o LGMD2C – SGCG:p.C283Y
- ◊ Muscle dystrophy panel (NGS)
  - o DYSF, CAPN3, SGCA, ANO5, SGCG, FKRP, DMD, POLG1 genes

Dystrophia Myotonica

- ◊ Dystrophia Myotonica type 1 (DMPK gene)
- ◊ Dystrophia Myotonica type 2 (ZNF9 gene)

Spinal Muscular Atrophy (SMA)

- ◊ SMN1 carrier screening
- ◊ SMA diagnostics (SMN1; SMN2, if positive)
- ◊ SMN2 (and SMN1) copy number examination (for therapy)
- ◊ SMN sequencing (when compound heterozygous state is suspected)

ALS (Amyotrophic Lateral Sclerosis)

- ◊ C9ORF72 gene repeat number determination
- ◊ SOD1 gene sequencing
- ◊ ALS panel (NGS)
  - o TARDP43, FUS, OPTN genes

Hereditary Neuropathies, Roma Founder Mutation

- ◊ Congenital cataract facial dysmorphia neuropathy CTDP1: c.863+389C>T
- ◊ Lom-NDRG1: c.442 C>T X-Linked CMT

CMT – Demyelination form

- ◊ copy number examination (MLPA):
  - o PMP22 (CMT1A/HNPP), CX32 genes
- ◊ PMP22 gene sequencing
- ◊ CX32 gene sequencing
- ◊ MPZ gene sequencing
- ◊ TTR gene sequencing

CMT2 – Axonal form

- ◊ CMT2 panel (NGS)
  - o PMP22, MFN2, MPZ, TTR, CX32, TTR, GDAP1, SH3TC2 genes

Myoglobinuria

- ◊ CPT-II gene sequencing

Congenital Myasthenia Syndrome

- ◊ CHRNE gene founder mutation analysis

Ophthalmological disorders

- ◊ LHON
- ◊ Norrie disease (NDP gene)
- ◊ RS1 gene sequencing
- ◊ DNAJC30 gene sequencing

Pulmonological disorders

- ◊ Alpha1 Antitrypsin deficiency (SERPINA1) gene sequencing

SYNDROMATIC PANELS

Marfan aortopathy Syndrome

- ◊ Panel sequencing (NGS)
  - o ACTA2, MYLK, ACTG2, TGFB2, COL3A1, FBN1, TGFBR1, TGFBR2, SMAD3, KCNN1, MYH11 genes

Neurocutaneous Syndromes

- ◊ Copy number examination (MLPA)
  - o NF1, NF2 genes
- ◊ Panel sequencing (NGS)
  - o GNAQ, KIT, LZTR1, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1, TSC1, TSC2 genes

SEGREGATIONS

- ◊ Segregation examination:

Name of gene for segregation .....
HGVS Nucleotide (c.): .....
HGVS Protein (p.): .....
Transcript ID (NM_): .....
Genome position (chr): .....
Name of Relative: .....
Degree of relation: .....

PANELS THAT MAY BE REQUESTED BASED ON INDIVIDUAL EQUITY

- ◊ **WES (Whole Exome Sequencing))**
- ◊ **Leucodystrophy panel**
- ◊ **Epilepsy panel**

We cannot examine samples arriving with an incomplete Genetic Test Request form until the missing information is provided.

Date:

Signature of doctor **l.s.**