

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
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Diseases of the Central Nervous System

Hereditary arteriopathy

- ◇ Hot spot analysis
 - NOTCH3 gene exon 3-4 (CADASIL)
- ◇ Arteriopathy panel (NGS)
 - NOTCH3, CTSA, HTRA1, COL4A1 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.
 - SCA1 (ATXN1 gene)
 - SCA2 (ATXN2 gene)
 - SCA3 (ATXN3 gene)
 - SCA6 (CACNA1 gene)
 - SCA7 (ATXN7 gene)
 - SCA17 (TRP gene)
- ◇ Friedreich ataxia - FXN gene repeat number det.
- ◇ Late Onset Ataxias repeat number det.
 - SCA27B (FGF14 gene)
 - CANVAS (RFC1 gene)
- ◇ Ataxia panel (NGS)
 - ATM, SACS, SETX, ANO10, APTX, SYNE1, CACNA1A, COQ4, COQ8A genes
- ◇ POLG1 gene sequencing (SANDO)

Parkinson's disease (familial or late onset)

- ◇ Copy number examination (MLPA)
 - SNCA, PARK2, PINK1, PARK7, ATP13A2, LRRK2 genes
- ◇ Hereditary Parkinson's disease panel (NGS)
 - 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)
 - PANK2, PLA2G6 genes
 - NBIA panel (NGS)
 - 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)
 - GRN, TARDBP43, MAPT genes
- ◇ Monogenic Alzheimer panel (NGS)
 - 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 (ACADMMD)

- ◇ 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)
 - POLG1, OPA1, TWNK, RRM2B, ANT1, SCO2, TK2 genes

Hereditary Spastic Paraparesis (HSP)

- ◇ Copy number examination (MLPA)
 - ATL1 (SPG3A), SPAST (SPG4) genes
- ◇ HSP panel (NGS)
 - SPAST (SPG4), SPG7, SPG11, AP4M1 genes

Intellectual Impairment/Autism Spectrum Disorder

- ◇ Copy number examination (MLPA)
 - Microdeletion MLPA (P245)
 - 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:
 - LGMD2C – SGCG:p.C283Y
- ◇ Muscle dystrophy panel (NGS)
 - DYSF, CAPN3, SGCA, ANO5, SGCG, FKRP, DMD, POLG1 genes

Dystrophia Myotonica

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

Oculopharyngeal muscular dystrophy (OPMD)

- ◇ PABPN1 gene repeat number det.

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)
 - TARDP43, FUS, OPTN genes

Hereditary Neuropathies, Roma Founder Mutation

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

CMT – Demyelination form

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

CMT2 – Axonal form

- ◇ CMT2 panel (NGS)
 - 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)
 - ACTA2, MYLK, ACTG2, TGFB2, COL3A1, FBN1, TGFBR1, TGFBR2, SMAD3, KCNN1, MYH11 genes

Neurocutaneous Syndromes

- ◇ Copy number examination (MLPA)
 - NF1, NF2 genes
- ◇ Panel sequencing (NGS)
 - 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

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