Request for Molecular Genetic Testing

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Head: Prof. Guntram Borck; MD, Human Genetics Specialist

PATIENT IDENTIFICATION (Label)

Surname _____ Prename _____

DOB _____ Gender (M/F) _____ Reference _____

Address _____

ZIP _____ City/Country _____

SENDER IDENTIFICATION
Contact Person (print) **Email:**

Sent Report to: (complete address)

Date _____ **Signature** _____

- ❑ E112 form appended
- ❑ Send invoice to: _____ (Billing address)
- ❑ ***You must check one box***

Material to be tested

- ❑ Blood sample, EDTA, 10 ml, freshly drawn, sent by overnight express service
- ❑ DNA sample: Sample volume (µl)*: _____, DNA concentration (µg/ml)* _____

If detection and characterization of disease alleles needs Southern blotting (e.g. fragile X, myotonic dystrophies, ALS) at least 15 µg at a minimum concentration of 200 µg/ml is required, which is usually not achieved by column extraction procedures..

- ❑ Other sample (*accepted only if previously announced*): _____

Indication for testing or questions to be answered: _____

Family history (clinical reports/pedigree appreciated. Use extra page if necessary)

Family member previously tested (in our lab): _____ (Name or reference (D number)) _____ (Relation to actual patient)

Further comments (e.g. week of gestation)

Test(s) requested for *(You must check at least one box)*:

Mental Retardation
- ❑ Angelman Syndrome (AS; *SNRPN*, ❑ *UBE3A*)
- ❑ ARX-associated mental Retardation (ARX)
- ❑ Börjeson-Forssman-Lehmann Syndrome (BFLS; *PHF6*)
- ❑ DiGeorge Syndrome (MLPA)
- ❑ Fragile-X Syndrome (*FMR1*) ❑ fragile X-associated tremor/ataxia syndrome (FXTAS)
- ❑ Miller-Dieker Lissencephaly (MLPA)
- ❑ Phelan-McDermid Syndrome // 22q13.3 Micro Deletion (*SHANK3*; MLPA, Sequencing)
- ❑ Prader-Willi Syndrome (PWS; *SNRPN*)
- ❑ Rett Syndrome (RTT; *MECP2*)
- ❑ Smith-Magenis Syndrome (MLPA)
- ❑ Williams-Beuren Syndrome (MLPA)

Neurological and neuromuscular disorders
- ❑ Amyotrophic lateral sclerosis, familial (❑ALS1[*SOD1*];❑ALS8[*VAPB*];❑ALS6[*FUS*]; ❑ALS10[*TARDBP=TDP43*]) ❑ ALS/FTD [*C9ORF72*]; PCR + Southern Blot)
- ❑ Alzheimer disease (familiäl) (❑ *PSEN1*, ❑ *PSEN2*, ❑ *APP*, ❑ *PRNP*)
- ❑ Chorea Huntington (❑ *HD*, Huntingtin; ❑ HDL2; *JPH3*) ❑ diagnostic ❑ predictive
- ❑ Creutzfeldt-Jakob Disease (CJD; *PRNP*)
- ❑ dentatorubral-pallidoluysian atrophy (*ATN1*)
- ❑ Fatal Familial Insomnia (FFI; *PRNP*)
- ❑ Friedreich Ataxia (FRDA1; Frataxin; *FXN*)
- ❑ Frontotemporale Demenz (❑ FTDP17; *MAPT*, ❑ HDDD; *GRN*) ❑ FTD/ALS[*C9ORF72*]; PCR + Southern Blot)
- ❑ Gerstmann-Sträussler Disease (GSD; *PRNP*)
- ❑ Muscular Dystrophy Duchenne/Becker (DMD/BMD; *DMD*)
- ❑ Myoclonic Epilepsy of Unverricht and Lundborg (EPM1; *CSTB*)
- ❑ Myotonic Dystrophy Type 1(DM1; *DMPK*)
- ❑ Myotonic Dystrophy Type 2 (DM2; ZNF9/CNBP)

- ❑ Oculopharyngeal Muscular Dystrophy (OPMD; *PABPN1*)
- ❑ Spinal and Bulbar Muscular Atrophy (SBMA/SMAX1; *AR*)
- ❑ Spinal Muscular Atrophy (SMA1-3; *SMN1*)
- ❑ Spinocerebellar Ataxia (all types listed below)
  - ❑ SCA1 (*ATXN1*) ❑ SCA6 (*CACNA1A*) ❑ SCA12 (*PPP2R2B*)
  - ❑ SCA2 (*ATXN2*) ❑ SCA7 (*ATXN7*) ❑ SCA14 (*PRKCG*)
  - ❑ SCA3/MJD (*ATXN3*) ❑ SCA8 (*ATXN8*) ❑ SCA17 (*TBP*)

Oncologic and Tumor-associated disorders
- ❑ Adenomatous Polyposis of the Colon (FAP; *APC*)
- ❑ Breast Cancer, familial ❑*BRCA1* ❑*BRCA2* ❑*RAD51C* ❑*RAD51D* ❑*CHEK2*
- ❑ Colorectal Adenomatous Polyposis, autosomal recessive (*MUTYH*)
- ❑ HNPCC (Lynch-Syndrome) (*MLH1, MSH2, MSH6, PMS2*)
- ❑ MAP (*MUTYH*)
- ❑ Multiple endocrine neoplasia MEN2A/B (*RET*)

Deafness
- ❑ Branchio-Oculo-Facial Syndrome (BOF; *TFAP2A*)
- ❑ Connexin 26 [*GJB2*] ❑ Connexin 30 [*GJB6*] = DFNB1
- ❑ KID syndrome (keratitis-ichthyosis-deafness) (*GJB2*)
- ❑ Pendred Syndrome (PDS; *SLC26A4 = PDS*)

Disorders of development and malformation syndromes
- ❑ Campomelic Dysplasia (*SOX9*)
- ❑ Adrenal Hypoplasia, congenital (AHC; *NR0B1/DAX1*)
- ❑ Swyer Syndrome (*SRY*, DSS-Region, *DMRT1*)
- ❑ XY Gonadal Dysgenesis (*SRY*, DSS-Region, *DMRT1, DHH*)

Pancreatitis
- ❑ Hereditary Pancreatitis
- ❑ cationic trypsinogen (*PRSS1*)
- ❑ Serine Protease Inhibitor, Kazal-Type 1 (*SPINK1*)
- ❑ Chymotrypsin C (*CTRC*)

Last change: 11/2014