Neuromuscular Research Department (NMRD) Vienna

We do care for muscle!

CENTER FOR ANATOMY AND CELL BIOLOGY
MEDICAL UNIVERSITY OF VIENNA

Patient data

Last Name: _____

First Name: _____

Date of Birth: _____ ☐ male ☐ female

Please enclose signed informed consent form!

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GENETIC TEST REQUISITION FORM

MUSCULAR DYSTROPHIES

- ☐ Duchenne/Becker muscular dystrophy (DMD/BMD)
- ☐ Facioscapulohumeral muscular dystrophy (FSHD)
- ☐ Emery-Dreifuss muscular dystrophy, X-linked (*EMD*)
- ☐ Emery-Dreifuss muscular dystrophy, X-linked, autos. (*LMNA*)
- ☐ Oculopharyngeal muscular dystrophy (OPMD)

Limb-girdle muscular dystrophies:

- ☐ LGMD1A (*MYOT*)
- ☐ LGMD1B (*LMNA*)
- ☐ LGMD1C (*CAV3*)
- ☐ LGMD2A (*CAPN3*)
- ☐ LGMD2B (*DYSF*)
- ☐ LGMD2C-E (*SGCG, SGCA, SGCB*)
- ☐ LGMD2I (*FKRP*)
- ☐ LGMD2K (*POMT1*)
- ☐ LGMD2L (*ANO5*)
- ☐ LGMD2M (*FKTN*)

CONGENITAL MUSCULAR DYSTROPHIES

- ☐ Rigid spine muscular dystrophy (*SEPN1*)
- ☐ Bethlem myopathy (*COL6A1-3*)
- ☐ Ullrich congenital muscular dystrophy (*COL6A1-3*)
- ☐ MDC1A (*LAMA2*)
- ☐ MDC1C (*FKRP*)
- ☐ MDC1D (*LARGE*)
- ☐ Muscle-Eye-Brain disease (*FKRP*, *POMGNT1*, *POMT1-2*)
- ☐ Walker-Warburg Syndrome (*FKRP*, *POMGNT1*, *POMT1-2*)

CONGENITAL MYOPATHIES

- ☐ Myotubular myopathy 1, X-linked. (*MTM1*)
- ☐ Myotubular myopathy, autos. (*DNM2, BIN1*)
- ☐ Central core disease (*RYR1*)
- ☐ Multi-Mini-Core disease (*SEPN1*, *RYR1*)
- ☐ Nemaline myopathy 3 (*ACTA1*)
- ☐ Myopathy, with fiber-type disproportion (*ACTA1*, *SEPN1*)
- ☐ Myofibrillar myopathy, ZASP-related (*LDB3*)
- ☐ Early onset myopathy, areflexia, resp. distress & dysphagia

DISTAL MYOPATHIES

- ☐ Miyoshi myopathy 1 (*DYSF*)
- ☐ Miyoshi myopathy 3 (*ANO5*)
- ☐ Distal myoptahy, with anterior tibial onset (*DYSF*)
- ☐ Distal myopathy 1, Laing (*MYH7*)
- ☐ Tardive tibial muscular dystrophy, Udd myopathy (*TTN*)

METABOLIC MYOPATHIES

- ☐ Glycogen storage disease Type II, V (*GAA*, *PYGM*)
- ☐ Myoadenylate deaminase deficiency (*AMPD1*)
- ☐ Carnitine palmitoyl-transferase deficiency (*CPT2*)

OTHER MYOPATHIES

- ☐ Desmin-related myofibrillar myopathy (*DES*, *SEPN1*)
- ☐ Danon disease (*LAMP2*)
- ☐ Scapuloperoneal myopathy, X-linked (*FHL1*)
- ☐ Inclusion body myopathy IBMPFD (*VCP*)

MYOTONIC SYNDROMES

- ☐ Myotonic dystrophy, type 1, Steinert (DM1)
- ☐ Myotonic dystrophy, type 2 (DM2, PROMM)
- ☐ Rippling muscle disease (*CAV3, PTRF, KCNA1*)

CONGENITAL MYASTHENIC SYNDROMES

- ☐ Multiple pterygium syndrome (*CHRNG*)
- ☐ Paramyotonia congenita (*SCN4A*)
- ☐ Hypo-/Hyperkalemic periodic paralysis (*SCN4A*)
- ☐ Myotonia congenita (*CLCN1*)
- ☐ Myasthenic syndrome, congenital (*RAPSN*)

SPINAL MUSCULAR ATROPHIES & MOTOR NEURON DISEASES

- ☐ Spinal muscular atrophy, type I-IV (*SMN1*)
- ☐ Spinal muscular atrophy with respiratory distress (SMARD1)
- ☐ Spinal and bulbar muscular atrophy, X-linked 1, Kennedy (*AR*)
- ☐ Amyotrophic lateral sclerosis (*SOD1, ALS2, VABP, VCP*)

DISORDERS OF NEURONAL MIGRATION

- ☐ Lissencephaly, X-linked (*DCX*)
- ☐ Polymicrogyria, bilateral frontoparietal (*GPR56*)

HEREDITARY MOTOR AND SENSORY NEUROPATHIES

- ☐ Hereditary neuropathy with pressure palsies, HNPP
- ☐ Charcot-Marie-Tooth disease (*PMP22, MFN2, GDAP1*)
- ☐ Charcot-Marie-Tooth disease, axonal, type 2B1 (*LMNA*)
- ☐ Charcot-Marie-Tooth neuropathy, X-linked (*GJB1*)
- ☐ Giant axonal neuropathy (*GAN1*)

NEURODEGENERATIVE LYSOSOMAL STORAGE DISEASES

- ☐ Ceroid lipofuscinosis, neuronal, type 1, CLN1 (*PPT1*)
- ☐ Ceroid lipofuscinosis, neuronal, type 2, CLN2 (*TPP1*)
- ☐ Ceroid lipofuscinosis, neuronal, type 10, CLN10 (*CTSD*)

DISORDERS OF OPTIC NERVE AND RETINA

- □ Stargardt disease1 (*ABCA4*, *ELOVL4*)
- □ Cone-rod dystrophy (*ABCA4, CRX*, *GUCY2D*)
- □ Leber Congenital amaurosis (*GUCY2D*, *RPE65, AIPL1, CRB1*)
- □ Macular dystrophy, vitelliform (*BEST1*)
- □ Retinitis pigmentosa (*RP1*, *RHO*, *PRPH2, RPGR, PRPF31, CRB1*)
- □ Basal laminar drusen (*EFEMP1, CFH*)
- □ Usher Syndrome, type 1B (*MYO7A*)
- □ Usher Syndrome, type 2A (*USH2A*)
- □ Usher Syndrome, type 2C (*GPR98*)
- □ Usher Syndrome, type 2D (*DFNB31*)
- □ Retinoschisis 1, juvenile, X-linked (*RS1*)
- □ Optic atrophy (*OPA1, OPA3, DNM1L, TMEM126A, ACO2, RTN4IP1, YME1L1, FDXR, SLC25A46, WFS1*)
- □ Achromatopsia (*CNGA3, CNGB3*)
- □ Albinism, ocular type (*OA1*)
- □ Albinism, oculocutaneous type 1 (*TYR*)
- □ Albinism, oculocutaneous type 2 (*OCA2*)
- □ Leber Hereditary Optic Neuropathy (mitochondrial genome)
- □ Wolfram Syndrome 1 (*WFS1*)
- □ Juvenile macular dystrophy, with hypotrichosis (*CDH3*)
- □ Sorsby fundus dystrophy (*TIMP3*)
- □ Microphthalmia, isolated 5 (*MFRP*)

MITOCHONDRIAL DISORDERS

- □ Progressive external ophthalmoplegia, autos. (*POLG*)
- □ Alpers Syndrome (*POLG*)
- □ mtDNA depletion syndrome, encephalomyopathic (*RRM2B*)
- □ mtDNA depletion syndrome, myopathic form (*TK2*)
- □ mtDNA depletion syndrome, hepatocerebral form (*DGUOK*)
- □ mtDNA depletion syndrome, MNGIE type (*TYMP*)
- □ Pyruvate dehydrogenase E3-binding protein deficiency (*PDHX*)
- □ Cardioencephalomyopathy, fatal infantile (*SCO2*)
- □ Progressive external ophthalmoplegia, autos. dom. (*TWINKLE*)
- □ Wolfram Syndrome 1 (*WFS1*)

NGS analysis of the mitochondrial genome (mtDNA):

- □ Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like episodes (MELAS)
- □ Myoclonic Epilepsy and Ragged Red Muscle Fibers (MERFF)
- □ Progressive Encephalopathy (PEM)
- □ Epilepsy, Strokes, Optic atrophy, & Cognitive decline (ESOC)
- □ Maternally Inherited Leigh Syndrome (MILS)
- □ Kearns-Sayre Syndrome (KSS)
- □ Pearson Syndrome
- □ Leber Hereditary Optic Neuropathy (LHON)
- □ Leber's hereditary optic neuropathy and Dystonia (LDYT)
- □ Progressive Dystonia
- □ Mitochondrial myopathy (MM)
- □ Maternally Inherited Diabetes and Deafness (MIDD)
- □ Wolfram Syndrome, mitochondrial form (DIDMOAD)
- □ Diabetes Mellitus & Deafness (DMDF)
- □ Maternally inherited deafness (DEAF)
- □ Sensorineural Hearing Loss (SNHL)
- □ Chronic Progressive External Ophthalmoplegia (CPEO)
- □ Maternal Myopathy and Cardiomyopathy (MMC)
- □ Maternally Inherited Cardiomyopathy (MICM)
- □ Familial Bilateral Striatal Necrosis (FBSN)
- □ Neurogenic muscle weakness, Ataxia, and Retinitis Pigmentosa (NARP)
- □ Ataxia, Myoclonus and Deafness (AMDF)

HyperCKemia

- □ Idiopathic hyperCKaemia (*CAV3*)
- □ Recurrent acute myoglobinuria, autos. rec. (*LPIN1*)
- □ other (*DYSF*, *CAPN3*)

HEREDITARY CARDIOMYOPATHIES

- □ CMD1A (*LMNA*)
- □ CMD1C (*LDB3*)
- □ CMD1G (*TTN*)
- □ CMD1I (*DES*)
- □ CMD3B, X-chr. (*DMD*)
- □ CMH (*CAV3*)
- □ CMH1 (*MYH7*)