THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
NEUROGENETICS DEPARTMENT
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NEUROGENETICS DEPARTMENT - REQUEST FOR DNA DIAGNOSTIC TESTS - Department Code: 28

Patient Information *(please complete accordingly)*

CASE TYPE: ☐ Outpatient ☐ Inpatient			Address
Name	**Surname**	Gender Male☐ Female ☐	
I.D. No.	D.O.B. / /	Nationality	Code City
CING No.	Family No.	Relation to proband	Country Telephone

Patient Status:

☐ GESY | ☐ Government-Non GESY: Hospital Card No. | ☐ Private-Non GESY

Requesting Clinician / Scientist

Name Surname Hospital / Clinic

Referring Clinician Status:

☐CING ☐Government (ΟΚΥπΥ) ☐Private-GESY GESY No.: ☐Private-Non GESY

Address

Code City

Telephone e-mail Signature

Diagnosis:

Date / /

The referring physician undertakes and confirms understanding and compliance in respect of the mutual obligations as these are determined under the GDPR.

Indication for Testing *(please complete accordingly)*

☐ Confirmation / exclusion of diagnosis ☐ Presymptomatic testing ☐ Carrier testing ☐ Other (please specify)
☐ Research ☐ Clinical Study ☐ Prenatal

Type of Specimen *(please complete accordingly)*

☐ Whole Blood ☐ Extracted DNA ☐ Biopsy ☐ CVS (Direct)
☐ CVS (Cultured) ☐ Amniotic fluid (Cultured) ☐ Amniotic Fluid (Direct)

Date Specimen collected / /

Sampling and transportation:

- Name, surname and date of birth should be clearly written
- 2 ml of blood in an EDTA tube is required for each individual **(For pre-symptomatic molecular diagnostic testing 2x vials for each individual are required)**
- Blood samples may be stored at room temperature or in the refrigerator until transport **(DO NOT FREEZE SAMPLES)**
- Blood samples should be transported by courier at room temperature and arrive within 72 hours (on Fridays before 13:00 hours)

Send samples to: **Kyproula Christodoulou, PhD**
Neurogenetics Department, The Cyprus Institute of Neurology and Genetics, 6, Iroon Avenue, Ayios Dhometios, 2371, Nicosia, Cyprus

CLINICAL FEATURES CHECKLIST *(please complete accordingly)*

Neurological / Neuromuscular
- ☐ Abnormality of the nervous system
- ☐ Abnormal movements
- ☐ Anosmia, congenital
- ☐ Ataxia
- ☐ Cerebral palsy
- ☐ Chorea
- ☐ Cognitive dysfunction
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Familial hemiplegic migraine
- ☐ Sporadic hemiplegic migraine
- ☐ Headaches
- ☐ Hypotonia
- ☐ Infantile spasms
- ☐ Peripheral neuropathy
- ☐ Psychiatric
- ☐ Memory impairment
- ☐ Muscle weakness
- ☐ Muscular dystrophy
- ☐ Nystagmus
- ☐ Seizures
- ☐ Sensory neuropathy
- ☐ Spasticity/ Hyperreflexia
- ☐ Stroke-like episode(s)
- ☐ Tremors

Cardiac
- ☐ Abnormal heart morphology
- ☐ Aortic root dilation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Cardiomyopathy ☐ DCM ☐ HCM ☐ Other
- ☐ Coarctation of aorta
- ☐ Heart murmur
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Patent ductus arteriosus
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect

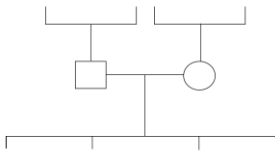
Ophthalmological
- ☐ Abnormal eye movement
- ☐ Abnormal vision
- ☐ Blindness
- ☐ Cataracts
- ☐ Coloboma
- ☐ PEO
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinitis pigmentosa

Development, Physical & Cognitive
- ☐ Delayed motor milestones
- ☐ Developmental regression
- ☐ Intellectual disability
- ☐ Speech/Language delay

Other Features
- ☐ Cancer/tumor formation
- ☐ Other skin findings
- ☐ Organomegaly
- ☐ Neuromas
- ☐ Pigmentary abnormalities

Additional clinical findings:

Pedigree: *(please complete and indicate the index case with an arrow)*



Patient History

Has the patient received a hematopoietic stem cell transplantation?
☐Yes ☐ No
Has the patient received granulocyte transfusions in the past two weeks?
☐Yes ☐ No
High infection risk: ☐Yes ☐ No
Clinical report enclosed: ☐Yes ☐No
Ethnic origin:

The Cyprus Institute of Neurology and Genetics (CING) complies with the General Data Protection Regulation EE 2016/679. For further information on how we protect personal data and on how data subjects can exercise their rights please refer to our privacy policy at https://www.cing.ac.cy/ or contact the CING Data Protection Officer at 22358600 or through the e-mail: dpo@cing.ac.cy.

Sample Receipt *(For Laboratory Internal Use)*

Received by	Signature	Sample Receipt Date:	DNA#
Amount	Comments:		

NEUROGENETICS DEPARTMENT - REQUEST FOR DNA DIAGNOSTIC TESTS

Patient Name:	***D.O.B.:*** / /

Test Required (Code No.) *(please complete accordingly)*

GENE DIAGNOSTICS WITH SEQ, MLPA, REPEAT ANALYSIS

	Code No
Amyloidosis:	
☐ Transthyretin Val30Met mutation detection test [FAP]*	1
☐ Transthyretin DNA sequencing test [FAP]*	1.01
Huntington Disease:	
☐ Huntington CAG triplet repeat test [HD]*	2
Ataxia:	
☐ Friedreich's Ataxia, Frataxin GAA triplet repeat test [FRDA]*	3
☐ SCA Panel (SCA1, 2, 3, 6, 7) test *	15
☐ Spinocerebellar Ataxia 1 (SCA1) CAG triplet repeat test [SCA1]*	4
☐ Spinocerebellar Ataxia 2 (SCA2) CAG triplet repeat test [SCA2]*	12
☐ Spinocerebellar Ataxia 3 (SCA3) CAG triplet repeat test [SCA3]*	5
☐ Spinocerebellar Ataxia 6 (SCA6) CAG triplet repeat test [SCA6]*	13
☐ Spinocerebellar Ataxia 7 (SCA7) CAG triplet repeat test [SCA7]*	14
☐ Spinocerebellar Ataxia 8 (SCA8) CTA/CTG repeat test [SCA8]	16
☐ Spinocerebellar Ataxia 10 (SCA10) ATTCT repeat test [SCA10]	21
☐ Spinocerebellar Ataxia 12 (SCA12) CAG triplet repeat test [SCA12]	17
☐ Spinocerebellar Ataxia 17 (SCA17) CAG/CAA repeat test [SCA17]	18
☐ DRPLA CAG triplet repeat test [DRPLA]	19
☐ Aprataxin (*APTX*) sequencing test [AOA1]*	20
☐ *RFC1* gene AAGGG pentanucleotide repeat test [CANVAS and other *RFC1*-related disorders]^{P}	72
☐ *FGF14* gene GAA triplet repeat expansion test [SCA27B]^{P}	73
Charcot-Marie-Tooth (CMT) disease – Demyelinating/ - Axonal and Hereditary neuropathy with liability to pressure palsies (HNPP):	
☐ Myelin Protein Zero *(MPZ)* sequencing test [CMT1B]*/ [CMT2I, CMT2J]*	6.02
☐ Connexin 32 *(CX32 / GJB1)* sequencing test [CMTX1, CX32]*	6.03
☐ Peripheral Myelin Protein 22 *(PMP22) sequencing test* [CMT1E]*/ [HNPP]	6.04
☐ CMT1A/HNPP MLPA evaluation [CMT1A]*/ [HNPP]	6.05
☐ Mitofusin 2 *(MFN2)* gene sequencing test [CMT2A]*	6.06
☐ Neurofilament-light (*NEFL*) gene sequencing test [CMT1F]*/ [CMT2E]*	6.07
☐ Ganglioside-induced differentiation-associated protein 1 (*GDAP1*) [CMT4A]*/ [CMT2K]*	6.08
☐ Glycyl-tRNA synthetase *(GARS)* gene sequencing test [CMT2D]*	6.09
☐ Early growth response 2 (*EGR2*) gene sequencing test [CMT1D, CMT4E]*	6.10
☐ Detection of the c.892C>T mutation in exon 5 of the LMNA gene [ARCMT2]*	6.11
Spinal Muscular Atrophy:	
☐ SMA MLPA evaluation [SMA1, SMA2, SMA3]*	7.01
☐ Spinal and bulbar muscular atrophy (SBMA) or Kennedy's disease, androgen receptor CAG triplet repeat test [SBMA, Kennedy]	22
☐ Glycyl-tRNA synthetase (*GARS*) gene sequencing test [DSMAV]*	6.09
☐ *BSCL2* gene targeted variant detection test (N88S&S90L) [HMN5]*	24
Muscular Dystrophy:	
☐ *DMD* gene MLPA evaluation [DMD/BMD]	71
☐ Known LGMD2 mutation detection test [LGMD]	8
Myotonic Dystrophy:	
☐ DM1 CTG triplet repeat test [DM1]	9
Amyotrophic Lateral Sclerosis (ALS)/ Frontotemporal dementia:	
☐ *SOD1* gene sequencing test [ALS1]*	23
☐ TAR DNA binding protein *TARDBP* (TDP-43) gene sequencing test [ALS10]*	25
☐ Fused in sarcoma (*FUS*) gene sequencing test [ALS6]^{P}*	32
☐ *C9orf72* gene GGGGCC hexanucleotide repeat test [ALSFTD]	33
Parkinson Disease:	
☐ Detection of the G2019S mutation in exon 41 of the *LRRK2* gene [PARK8]	26
Hereditary Spastic Paraplegia:	
☐ Gap junction protein, gamma 2 (*GJC2*) gene sequencing test [SPG44]*	27
☐ Spastin (*SPAST*) gene sequencing test [SPG4]*	28
☐ Atlastin GTPase 1 (*ATL1*) gene sequencing test [SPG3]*	29
☐ *SPAST* and *ATL1* genes MLPA evaluation [SPG3] [SPG3A]*	30
☐ Receptor expression enhancing protein 1 (*REEP1*) gene sequencing test [SPG31]^{P}*	31
Cardiomyopathy:	
☐ Lamin A/C (*LMNA*) gene sequencing test [CMD1A]^{P}*	34
☐ Molecular investigations of Cardiomyopathies [HCM, DCM]	37
Migraine:	
☐ ATPase, Na+/K+ transporting, alpha 2 polypeptide (*ATP1A2)* gene sequencing test [FHM2]^{P}	35
Osteoporosis:	
☐ Low density lipoprotein receptor-related protein 5 gene sequencing test (*LRP5*)^{P}	36
Family Analysis:	
☐ Specify disease & locus: _____	80
Prenatal Diagnosis:	
☐ Prenatal diagnosis 1st specify disease, locus & variant: _____	81
☐ Prenatal diagnosis 2nd or later specify disease, locus & variant: _____	82
Other specific test:	
☐ Analysis of known gene mutation for additional family members- specify disease, locus & variant: _____	83
Other:	
☐ DNA extraction & banking*	10

NGS PANELS/ ADVANCED EVALUATION

	Code No
Please check the box if diagnostic NGS tests will be requested and fill in the following fields: ☐ Current NGS request is for diagnostic purposes only. Please state if any other diagnostic tests have been performed for this patient **Yes/ No** If **Yes** state which tests: _____ If **No** state why patient is referred for NGS as a first -tier test: _____	
☐ Amyotrophic Lateral Sclerosis panel*	41
☐ Ataxia panel*	42
☐ Cardiomyopathy panel*	40.2
☐ Cerebral Cavernous Malformation panel*	43
☐ Charcot-Marie-Tooth Neuropathy panel*	44
☐ Congenital Myasthenic Syndromes panel*	45
☐ Custom Neuro panel*	46
☐ Dementia panel*	47
☐ Distal Hereditary Motor Neuropathy panel*	48
☐ Dystonia panel*	49
☐ Epilepsy panel*	50
☐ Hemiplegia/Stroke panel*	51
☐ Leukodystrophy and Leukoencephalopathy panel*	52
☐ Migraine panel*	53
☐ Muscular Dystrophy-Myopathy panel*	54
☐ Neurofibromatosis panel*	55
☐ Neuro-ophthalmic panel*	56
☐ Parkinson Disease panel*	57
☐ Retinal degeneration panel*	58
☐ Spastic Paraplegia panel*	59
☐ Spinal Muscular Atrophy panel*	60
☐ Tuberous Sclerosis panel*	61
☐ Whole Exome Sequencing-WES (Single)*	64
☐ Whole Exome Sequencing-WES (Trio)*	65
☐ Open WES data after in silico panel*	67
☐ Trio in silico panel from WES (for all above disease specific panels)*	69
☐ Sanger sequencing-based confirmation of a variant identified by NGS*	70

ABBREVIATIONS

MLPA: Multiplex ligation-dependent probe amplification
SEQ: Sanger Sequencing **WES:** Whole Exome Sequencing
P: Private only
*: Accredited according to ISO 15189:2012

New forms can be ordered (roula@cing.ac.cy) or downloaded from (http://www.cing.ac.cy/easyconsole.cfm/id/364)