THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

Kyproula Christodoulou, PhD, Neurogenetics Department
6 International Airport Avenue, Ayios Dhometios, 2370 NICOSIA, CYPRUS
TEL.: +357-22358600, +357-22392649, FAX: +357-22392615, e-mail: roula@cing.ac.cy

NEUROGENETICS DEPARTMENT - REQUEST FOR DNA DIAGNOSTIC TESTS

Department Code: 28

Patient Identification *(Please tick ☑ accordingly)*	*Requesting Clinician / Scientist*
Name:_____ Surname: _____	Name: _____ Surname: _____
Sex: ☐ Male ☐ Female Date of Birth: ____ /____ /____	Hospital / Clinic: _____
I.D. No.: _____ Nationality: _____	Address: _____
CING No.: _____ Hospital File No.: _____	City: _____ Code: _____ Country: _____
Hospital Card No.: _____ Patient Status: ☐GP ☐ PP	Phone: _____ Fax: _____
Address: _____	e-mail: _____
City: _____ Code: _____ Country: _____	Diagnosis:_____
Phone: Home: _____ Work: _____	_____
Family No.: _____ Relation to proband: _____	Signature: _____ Date: ____ / ____ / ____

Indication for Testing *(Please tick ☑ accordingly)* ☐ Confirmation / exclusion of diagnosis ☐ Presymptomatic testing ☐ Carrier testing ☐ Prenatal
☐ Research ☐ Clinical Study ☐ Other (please specify) _____

Type of Specimen *(Please tick ☑ accordingly)* ☐ Whole Blood ☐ Extracted DNA ☐ Muscle ☐ CVS (Direct) ☐ CVS (Cultured)
☐ Amniotic fluid (Cultured) ☐ Amniotic Fluid (Direct) ☐ Archival material (please specify) _____ Date specimen collected: ____ / ____ / ____

Test Required (Code No.) *(Please tick ☑ accordingly)*

Amyloidosis:
☐ Transthyretin (*TTR*) gene Val30Met mutation detection test [FAP] *(1)*
☐ Transthyretin (*TTR*) gene sequencing test [FAP] *(1.01)*

Huntington Disease:
☐ Huntingtin (*HTT*) gene CAG triplet repeat test [HD] *(2)*

Ataxia:
☐ Frataxin (*FXN*) gene GAA triplet repeat test [FRDA, Friedreich ataxia] *(3)*
☐ Ataxin 1 (*ATXN1*) gene CAG triplet repeat test [SCA1] *(4)*
☐ Ataxin 2 (*ATXN2*) gene CAG triplet repeat test [SCA2] *(12)*
☐ Ataxin 3 (*ATXN3*) gene CAG triplet repeat test [SCA3] *(5)*
☐ A1a voltage-dependent calcium channel subunit (*CACNA1A*) gene CAG triplet repeat test [SCA6] *(13)*
☐ Ataxin 7 (*ATXN7*) gene CAG triplet repeat test [SCA7] *(14)*
☐ SCA Panel (SCA1, 2, 3, 6, 7) test *(15)*
☐ Spinocerebellar Ataxia 8 CTA/CTG repeat test [SCA8] *(16)*
☐ Ataxin 10 (*ATXN10*) gene ATTCT repeat test [SCA10] *(21)*
☐ Protein phosphatase 2, regulatory subunit B (*PPP2R2B*) gene CAG triplet repeat test [SCA12] *(17)*
☐ TATA box binding protein (*TBP*) gene CAG/CAA repeat test [SCA17] *(18)*
☐ Atrophin 1 (*ATN1*) gene CAG triplet repeat test [DRPLA] *(19)*
☐ Aprataxin (*APTX*) gene sequencing test [AOA1] *(20)*

Charcot-Marie-Tooth (CMT) disease - Demyelinating:
☐ Peripheral myelin protein 22 (*PMP22*) gene dosage evaluation by MLPA analysis [CMT1A] *(6.05)*
☐ Myelin Protein Zero (*MPZ*) gene sequencing test [CMT1B] *(6.02)*
☐ Connexin 32 (*CX32 / GJB1*) gene sequencing test [CMTX1, CX32] *(6.03)*
☐ Peripheral Myelin Protein 22 (*PMP22*) gene sequencing test [CMT1E] *(6.04)*
☐ Neurofilament-light (*NEFL*) gene sequencing test [CMT1F] *(6.07)*
☐ Ganglioside-induced differentiation-associated protein 1 (*GDAP1*) gene sequencing test [CMT4A] *(6.08)*
☐ Early growth response 2 (*EGR2*) gene sequencing test [CMT1D, CMT4E] *(6.10)*

Charcot-Marie-Tooth (CMT) disease - Axonal:
☐ Mitofusin 2 (*MFN2*) gene sequencing test [CMT2A] *(6.06)*
☐ Connexin 32 (*CX32 / GJB1*) gene sequencing test [CMTX1, CX32] *(6.03)*
☐ Myelin Protein Zero (*MPZ*) gene sequencing test [CMT2I, CMT2J] *(6.02)*
☐ Glycyl-tRNA synthetase (*GARS*) gene sequencing test [CMT2D] *(6.09)*
☐ Neurofilament-light (*NEFL*) gene sequencing test [CMT2E] *(6.07)*
☐ Ganglioside-induced differentiation-associated protein 1 (*GDAP1*) gene sequencing test [CMT2K] *(6.08)*
☐ Detection of the c.892C>T mutation in exon 5 of the *LMNA* gene [ARCMT2] (*6.11)*

Hereditary neuropathy with liability to pressure palsies (HNPP):
☐ Peripheral myelin protein 22 (*PMP22*) gene dosage evaluation by MLPA analysis [HNPP] *(6.05)*
☐ Peripheral Myelin Protein 22 (*PMP22*) gene sequencing test [HNPP] *(6.04)*

Spinal Muscular Atrophy:
☐ Survival motor neuron 1 (*SMN1*) and 2 (*SMN2*) gene dosage evaluation by MLPA analysis [SMA1, SMA2, SMA3] *(7.01)*
☐ Androgen receptor (*AR*) gene CAG triplet repeat test [SBMA, Kennedy] *(22)*
☐ Glycyl-tRNA synthetase (*GARS*) gene sequencing test [DSMAV] *(6.09)*
☐ Detection of the N88S and S90L mutations in exon 3 of the *BSCL2* gene [HMN5]*(24)*

Myotonic Dystrophy:
☐ Dystrophia myotonica protein kinase (*DMPK*) gene CTG triplet repeat test [DM1] *(9)*

Amyotrophic Lateral Sclerosis (ALS):
☐ Superoxide dismutase 1 (*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] *(32)** [private only]

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 [SPG3A] *(29)*
☐ Spastin (*SPAST*) and Atlastin (*ATL1*) gene dosage evaluation by MLPA analysis [SPG4, SPG3A] *(30)*
☐ Receptor expression enhancing protein 1 (*REEP1*) gene sequencing test [SPG31] *(31)** [private only]

Other specific test:
☐ Disease:_____
Gene:_____ Family #:_____

Family Analysis:
☐ Please specify disease & locus *(80)*: _____

Prenatal Diagnosis:
☐ 1st - please specify disease, locus & mutation *(81)*: _____
☐ 2nd or later - specify disease, locus & mutation *(82)*: _____

Other:
☐ DNA extraction & banking *(10)*
☐ Research study *(For Laboratory Internal Use)*
Project: _____ Funding body: _____

⇒ Please, provide relevant information on the back of this form

Sample Receipt (*For Laboratory Internal Use)* Sample Receipt Date: ____ / ____ / ____

Received by: _____ Signature: _____
Amount: _____ Comments: _____ DNA #: _____

Patient Name: _____ Date of birth: ____ / ____ / ____

Comment / Clinical information:

Clinical report enclosed: ☐ Yes / ☐ No

Clinical questionnaire enclosed: ☐ Yes / ☐ No

_____

_____

_____

_____

_____

Ethnic origin: _____

Family information:

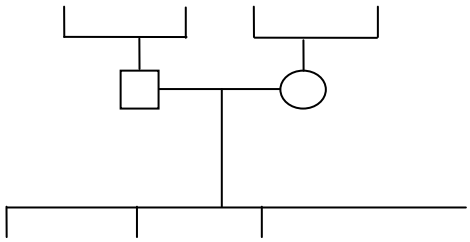
☐ Isolated patient

☐ Familial: ☐ dominant ☐ recessive ☐ X-linked

☐ Parents related : ☐ Yes / ☐ No If yes, please specify: _____

Pedigree: (please complete pedigree; indicate the index case with an arrow)

☐ Male
O Female
◊ Unknown gender
■ Affected male
● Affected female
⊠ Deceased male
Ø Deceased female



Sampling and transportation:

⇒ Name, surname and date of birth should be mentioned on each tube.
⇒ 2 ml of blood in EDTA tube is required for each individual.
⇒ Blood samples may be stored at room temperature or the refrigerator until transport (DO NOT FREEZE SAMPLES).
⇒ Blood samples should be transported by courier at room temperature and arrive within 72 hours (on Friday before 14:00 hours).
⇒ Send samples to: Kyproula Christodoulou, PhD,
Neurogenetics Department,
The Cyprus Institute of Neurology and Genetics,
6 International Airport Avenue,
Ayios Dhometios, 2370 Nicosia, Cyprus.

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