



THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

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NEUROGENETICS DEPARTMENT - REQUEST FOR DNA DIAGNOSTIC TESTS

Department Code: 28

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

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)

<p>Amyloidosis: <input type="checkbox"/> Transthyretin (<i>TTR</i>) gene Val30Met mutation detection test [FAP] (1) <input type="checkbox"/> Transthyretin (<i>TTR</i>) gene sequencing test [FAP] (1.01)</p> <p>Huntington Disease: <input type="checkbox"/> Huntingtin (<i>HTT</i>) gene CAG triplet repeat test [HD] (2)</p> <p>Ataxia: <input type="checkbox"/> Frataxin (<i>FXN</i>) gene GAA triplet repeat test [FRDA, Friedreich ataxia] (3) <input type="checkbox"/> Ataxin 1 (<i>ATXN1</i>) gene CAG triplet repeat test [SCA1] (4) <input type="checkbox"/> Ataxin 2 (<i>ATXN2</i>) gene CAG triplet repeat test [SCA2] (12) <input type="checkbox"/> Ataxin 3 (<i>ATXN3</i>) gene CAG triplet repeat test [SCA3] (5) <input type="checkbox"/> A1a voltage-dependent calcium channel subunit (<i>CACNA1A</i>) gene CAG triplet repeat test [SCA6] (13) <input type="checkbox"/> Ataxin 7 (<i>ATXN7</i>) gene CAG triplet repeat test [SCA7] (14) <input type="checkbox"/> SCA Panel (SCA1, 2, 3, 6, 7) test (15) <input type="checkbox"/> Spinocerebellar Ataxia 8 CTA/CTG repeat test [SCA8] (16) <input type="checkbox"/> Ataxin 10 (<i>ATXN10</i>) gene ATTCT repeat test [SCA10] (21) <input type="checkbox"/> Protein phosphatase 2, regulatory subunit B (<i>PPP2R2B</i>) gene CAG triplet repeat test [SCA12] (17) <input type="checkbox"/> TATA box binding protein (<i>TBP</i>) gene CAG/CAA repeat test [SCA17] (18) <input type="checkbox"/> Atrophin 1 (<i>ATN1</i>) gene CAG triplet repeat test [DRPLA] (19) <input type="checkbox"/> Aprataxin (<i>APTX</i>) gene sequencing test [AOA1] (20)</p> <p>Charcot-Marie-Tooth (CMT) disease - Demyelinating: <input type="checkbox"/> Peripheral myelin protein 22 (<i>PMP22</i>) gene dosage evaluation by MLPA analysis [CMT1A] (6.05) <input type="checkbox"/> Myelin Protein Zero (<i>MPZ</i>) gene sequencing test [CMT1B] (6.02) <input type="checkbox"/> Connexin 32 (<i>CX32 / GJB1</i>) gene sequencing test [CMTX1, CX32] (6.03) <input type="checkbox"/> Peripheral Myelin Protein 22 (<i>PMP22</i>) gene sequencing test [CMT1E] (6.04) <input type="checkbox"/> Neurofilament-light (<i>NEFL</i>) gene sequencing test [CMT1F] (6.07) <input type="checkbox"/> Ganglioside-induced differentiation-associated protein 1 (<i>GDAP1</i>) gene sequencing test [CMT4A] (6.08) <input type="checkbox"/> Early growth response 2 (<i>EGR2</i>) gene sequencing test [CMT1D, CMT4E] (6.10)</p> <p>Charcot-Marie-Tooth (CMT) disease - Axonal: <input type="checkbox"/> Mitofusin 2 (<i>MFN2</i>) gene sequencing test [CMT2A] (6.06) <input type="checkbox"/> Connexin 32 (<i>CX32 / GJB1</i>) gene sequencing test [CMTX1, CX32] (6.03) <input type="checkbox"/> Myelin Protein Zero (<i>MPZ</i>) gene sequencing test [CMT2I, CMT2J] (6.02) <input type="checkbox"/> Glycyl-tRNA synthetase (<i>GARS</i>) gene sequencing test [CMT2D] (6.09) <input type="checkbox"/> Neurofilament-light (<i>NEFL</i>) gene sequencing test [CMT2E] (6.07) <input type="checkbox"/> Ganglioside-induced differentiation-associated protein 1 (<i>GDAP1</i>) gene sequencing test [CMT2K] (6.08) <input type="checkbox"/> Detection of the c.892C>T mutation in exon 5 of the <i>LMNA</i> gene [ARCMT2] (6.11)</p>	<p>Hereditary neuropathy with liability to pressure palsies (HNPP): <input type="checkbox"/> Peripheral myelin protein 22 (<i>PMP22</i>) gene dosage evaluation by MLPA analysis [HNPP] (6.05) <input type="checkbox"/> Peripheral Myelin Protein 22 (<i>PMP22</i>) gene sequencing test [HNPP] (6.04)</p> <p>Spinal Muscular Atrophy: <input type="checkbox"/> Survival motor neuron 1 (<i>SMN1</i>) and 2 (<i>SMN2</i>) gene dosage evaluation by MLPA analysis [SMA1, SMA2, SMA3] (7.01) <input type="checkbox"/> Androgen receptor (<i>AR</i>) gene CAG triplet repeat test [SBMA, Kennedy] (22) <input type="checkbox"/> Glycyl-tRNA synthetase (<i>GARS</i>) gene sequencing test [DSMAV] (6.09) <input type="checkbox"/> Detection of the N88S and S90L mutations in exon 3 of the <i>BSCL2</i> gene [HMN5] (24)</p> <p>Myotonic Dystrophy: <input type="checkbox"/> Dystrophin myotonic protein kinase (<i>DMPK</i>) gene CTG triplet repeat test [DM1] (9)</p> <p>Amyotrophic Lateral Sclerosis (ALS): <input type="checkbox"/> Superoxide dismutase 1 (<i>SOD1</i>) gene sequencing test [ALS1] (23) <input type="checkbox"/> TAR DNA binding protein TARDBP (<i>TDP-43</i>) gene sequencing test [ALS10] (25) <input type="checkbox"/> Fused in sarcoma (<i>FUS</i>) gene sequencing test [ALS6] (32)* [private only]</p> <p>Parkinson Disease: <input type="checkbox"/> Detection of the G2019S mutation in exon 41 of the <i>LRRK2</i> gene [PARK8] (26)</p> <p>Hereditary Spastic Paraplegia: <input type="checkbox"/> Gap junction protein, gamma 2 (<i>GJC2</i>) gene sequencing test [SPG44] (27) <input type="checkbox"/> Spastin (<i>SPAST</i>) gene sequencing test [SPG4] (28) <input type="checkbox"/> Atlastin GTPase 1 (<i>ATL1</i>) gene sequencing test [SPG3A] (29) <input type="checkbox"/> Spastin (<i>SPAST</i>) and Atlastin (<i>ATL1</i>) gene dosage evaluation by MLPA analysis [SPG4, SPG3A] (30) <input type="checkbox"/> Receptor expression enhancing protein 1 (<i>REEP1</i>) gene sequencing test [SPG31] (31)* [private only]</p> <p>Other specific test: <input type="checkbox"/> Disease: _____ Gene: _____ Family #: _____</p> <p>Family Analysis: <input type="checkbox"/> Please specify disease & locus (80): _____</p> <p>Prenatal Diagnosis: <input type="checkbox"/> 1st - please specify disease, locus & mutation (81): _____ <input type="checkbox"/> 2nd or later - specify disease, locus & mutation (82): _____</p> <p>Other: <input type="checkbox"/> DNA extraction & banking (10) <input type="checkbox"/> Research study (For Laboratory Internal Use) Project: _____ Funding body: _____</p>
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⇒ Please, provide relevant information on the back of this form

Sample Receipt (For Laboratory Internal Use)

Sample Receipt Date: ____ / ____ / ____

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

Patient Name: _____

Date of birth: ____ / ____ / ____

Comment / Clinical information:

Clinical report enclosed: Yes / No

Clinical questionnaire enclosed: Yes / No

Ethnic origin: _____

Family information:

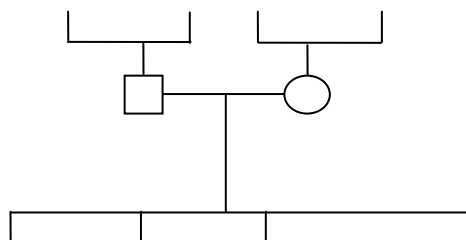
Isolated patient

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
- Female
- Unknown gender
- Affected male
- Affected female
- Deceased male
- 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: Kyroula 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>).