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NEUROGENETICS DEPARTMENT - REQUEST FOR DNA DIAGNOSTIC TESTS - Department Code: 28

Patient Information (please complete accordingly)											
CASE TYPE: Outpatient		□ Inpatient				Addres	s				
Name	Surn	•	Gen	ıder							
			Mal		Female 🗆						
I.D. No.	D.O.E	3.	Nati	ionality		Code		City			
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Patient Status:											
□ GESY □ Governm	nent-No	on GESY: Hospital Card No.						Private-Non GESY			
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Requesting Clinician / Scientist											
Name Suri	name		Hospital /	Clinic			Diagnos	is.			
	lanic		nospital /	Gilline			Diagnos	15.			
Referring Clinician Status:											
\Box CING \Box Government (OKY π Y)		Private-GESY GESY No.:			□ Private-Non	GESY					
Address											
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Telephone e-m	ail		8				Date	/ /			
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The referring physician undertakes and con	firms und	ierstanding and compliance in respe	ect of the mutu	al obligatio	ns as these are det	ermined ur	nder the GDI	РК.			
Indication for Testing (please compl	lete acco	ordingly)									
□ Confirmation / exclusion of diagno		□ Presymptomatic tes	sting	🗆 Carrie	er testing	🗆 Oth	ner (please	e specify)			
□ Research		□ Clinical Study	-	🗆 Prena	0		**				
	1	5		-							
Type of Specimen (please complete of Specimen)		01)					ata Cri	an collecte 3			
□ Whole Blood □ Extra		— P-J			🗆 CVS (Direct	;) Da	ate Specin	nen collected			
\Box CVS (Cultured) \Box Amni	otic flui	d (Cultured) 🛛 🗆 Amnioti	ic Fluid (Dire	ect)				/ /			
Sampling and transportation:											
 Name, surname and date of birth sh 	ould be	clearly written									
			mptomatic r	molecular	r diagnostic tes	ting 2x v	vials for early	ach individual are required			
 Blood samples may be stored at room 	m temp		 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 	ed by co	urier at room temperature and					00 hours)				
Send samples to: Kyproula Christon	loulou,	PhD	l arrive withi	in 72 hour	rs (on Fridays be	efore 13:(
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Patient Name:

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GENE DIAGNOSTICS WITH SEQ, MLPA, REPEAT ANALYSIS Code No Amvloidosis: □ 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 RFC1-72 related disorders] □ *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]*/ П 6.04 [HNPP] □ CMT1A/HNPP MLPA evaluation [CMT1A]*/ [HNPP] 6.05 □ Mitofusin 2 (MFN2) gene sequencing test [CMT2A]* 6.06 \square Neurofilament-light (NEFL) gene sequencing test [CMT1F]*/ [CMT2E]* 6.07 Ganglioside-induced differentiation-associated protein 1 (GDAP1) 6.08 [CMT4A]*/ [CMT2K]* 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 \square 6.11 [ARCMT2]* Spinal Muscular Atrophy: SMA MLPA evaluation [SMA1, SMA2, SMA3]* 7.01 Spinal and bulbar muscular atrophy (SBMA) or Kennedy's disease, \square 22 androgen receptor CAG triplet repeat test [SBMA, Kennedy] □ 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 25 [ALS10]* □ Fused in sarcoma (*FUS*) gene sequencing test [ALS6]^{P*} 32 C9orf72 gene GGGGCC hexanucleotide repeat test [ALSFTD] \square 33 Parkinson Disease: Detection of the G2019S mutation in exon 41 of the LRRK2 gene \square 26 [PARK8] 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 \square SPAST and ATL1 genes MLPA evaluation [SPG3] [SPG3A]* 30 Receptor expression enhancing protein 1 (REEP1) gene sequencing test 31 [SPG31]P*

Cardiomyopathy: Imamin A/C (LMAM) gene sequencing test [CMD1A]** 34 Molecular investigations of Cardiomyopathies [HCM, DCM] 37 Migraine: ATPase, Na+/K+ transporting, alpha 2 polypeptide (ATP1A2) gene sequencing test [FHM2]* 35 Osteoporosis: I.cow density lipoprotein receptor-related protein 5 gene sequencing test [(LR75)*)* 80 Prenatal Diagnosis: 90 90 Prenatal Diagnosis: 91 91 Prenatal diagnosis 1* 81 92 specify disease, locus & variant: 92 92 Other specific test: 91 91 92 Other specific test: 92 91 92 Other specific test: 91 91 91 DNA extraction & banking* 10 91 91 NGS PANELS/ ADVANCED EVALUATION Code NC 92 Vers of predict test: 11 11 92 If Ne state which tests: 11 11 94 If No state why patient is referred for NGS as a first-tier test: 11 14 14 Cardiomyopathy panel* 42 42 42 42 43 44 <td< th=""><th></th><th></th><th></th></td<>							
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New forms can be ordered (<u>roula@cing.ac.cy</u>) or downloaded from (http://www.cing.ac.cy/easyconsole.cfm/id/364)

F07.02.03.ND.01_Request form for DNA diagnostic tests_v1

Prepared by: AG, KC