THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

Referral Letter

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Email: mgftlab@cing.ac.cy
Department Code: 27

Department of Molecular Genetics, Function & Therapy
Head: Leonidas Phylactou PhD

DNA/MS/ST # _____ ***Dept. code*** _____

Patient details CING No.: _____ (if applicable)

Name: _____ Surname: _____

D.O.B.: _____/_____/_____ I.D. No.: _____

Nationality: _____ Gender: ☐Male ☐Female

Patient Status:

☐GESY ☐ Private-Non GESY

☐ Government-Non GESY Hospital Card No.: _____

Case type: ☐Outpatient ☐ Inpatient

Address: _____

City: _____ Code: _____ Country: _____

Phone: _____ e-mail: _____

Informed Consent

I authorize the Department of Molecular Diagnosis Function and Therapy to use my (or my child's/my foetus) sample (whole blood, serum or CSF) for test validation or research purposes after the personal details are removed making the procedure anonymous.

I have the right to refuse the above without having any consequence to the analysis. Samples will be stored for future reference or use.

- I can withdraw my consent at any time by contacting the laboratory at +35722392659.

Patient/Guardian Signature: _____ Date____/____/_____

Sample Receipt (*For Laboratory Internal Use*)

Received by: _____ **Signature**: _____

Receipt Date: ____/____/____ **Sample Type & Amount**:_____

Referring clinician's / scientist's details

Name: _____ Surname: _____

Hospital / Clinic: _____

Address: _____

City: _____ Code: _____ Country: _____

Phone: _____ Fax: _____

e-mail: _____

Reason for Referral: _____

Clinician Signature: _____ Date: _____/_____ /_____

Referring clinician status:

☐CING ☐Government (ΟΚΥΠΥ) ☐ Private -Non GESY

☐Private-GESY GESY No. _____

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

Please tick the box if diagnostic NGS tests are been requested and fill in the following fields: ☐ Current NGS referral 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: _____

_____

Sample details *(Please tick ☑ accordingly)*

Date and Time of Sample Collection: _____

Sample: ☐Blood (3-4ml) ☐CSF (0.5ml) ☐Urine ☐Muscle
☐CVS Other (please specify): _____
☐First Investigation ☐Repetition

For Genetic testing 3-4ml whole blood in EDTA is required.
For Multiple Sclerosis 0.5ml Serum and 0.5ml CSF are required.

Test Required *(Please tick ☑ accordingly)*

CFTR GENE, CYSTIC FIBROSIS (CF)

☐ *CFTR* analysis by NGS (1.1) *ISO 15189 ***(GESY)***

Sweat Test: ☐ Sweat test (8.7) ***(GESY)***

HEREDITARY RECURRENT FEVERS (HRFS)

☐ Hereditary recurrent fevers (HRFs) *in silico* panel from WES (24.3) *ISO 15189 ***(GESY)***

***MEFV* Gene, Familial Mediterranean Fever** *ISO 15189

☐ *MEFV* gene mutation analysis (sequencing Exons 2, 3, 5 & 10) (7.1) ***(GESY)***

☐ *MEFV* gene analysis for known mutation (sequencing) (7.2) ***(GESY)***

Mevalonate kinase Deficiency - MKD

☐ *MVK* gene full mutation analysis (sequencing) (18.1) (*Non-GESY*)

☐ *MVK* gene analysis for known mutation (sequencing) (18.2) (*Non-GESY*)

Tumour Necrosis factor associated periodic syndrome – TRAPS

☐ *TNFRSF1A* gene full mutation analysis (sequencing) (23.1) (*Non-GESY*)

☐ *TNFRSF1A* gene analysis for known mutation (sequencing) (23.2) (*Non-GESY*)

Cryopyrin-associated periodic syndrome – CAPS

☐ *NLRP3* gene full mutation analysis (sequencing) (24.1) (*Non-GESY*)

☐ *NLRP3* gene analysis for known mutation (sequencing) (24.2) (*Non-GESY*)

INHERITED DEAFNESS

☐ Hearing Loss *in silico* panel from WES (15.6) *ISO 15189 ***(GESY)***

Connexin 26 & 30, Inherited Deafness *ISO 15189

☐ Connexin 26 full mutation analysis (sequencing) (13.1) ***(GESY)***

☐ Connexin 26 analysis for known mutation (sequencing) (13.2) ***(GESY)***

☐ Connexin 30, Del(GJB6-D13S1830) (15.1) ***(GESY)***

HAEMOCHROMATOSIS

HFE* Gene, HaemochromatosisISO 15189

HFE gene, Haemochromatosis analysis of individual mutations (10.7) ***(GESY)***

☐ p.H63D ☐ p.C282Y

MULTIPLE SCLEROSIS

☐ Oligoclonal Bands detection for MS patients (14) *ISO 15189 ***(GESY)***

EXOME SEQUENCING *ISO 15189

☐ Whole Exome Sequencing (WES) (43) ***(GESY)***) / ☐ WES (Trio) (44) ***(GESY)***

☐ Sanger sequencing based confirmation of a variant identified by NGS (46) ***(GESY)***

DISORDERS OF SEXUAL DIFFERENTIATION

☐ Disorders of Sexual Development *in silico* panel from WES (45) *ISO 15189 ***(GESY)***

***CYP21A2* Gene, Congenital Adrenal Hyperplasia (CAH)** *ISO 15189

☐ *CYP21A2* gene full mutation analysis (sequencing & MLPA) (16.1) ***(GESY)***

☐ *CYP21A2* gene analysis for known mutation (sequencing) (16.2) ***(GESY)***

***SRD5α* Gene, 5α Reductase Deficiency, Disorder of Sexual Differentiation** *ISO 15189

☐ *SRD5A2* gene, full mutation analysis (sequencing) (21.1) (*Non-GESY*)

☐ *SRD5A2* gene analysis for known mutation (sequencing) (21.2) (*Non-GESY*)

***PROP1* Gene, Growth Hormone Deficiency**

☐ *PROP1* full mutation analysis (sequencing) (22.1) (*Non-GESY*)

☐ *PROP1* DNA extraction/storage (22.6) (*Non-GESY*)

PREMATURE AND DELAYED PUBERTY

☐ Premature and Delayed Puberty *in silico* panel from WES (38.4) *ISO 15189 ***(GESY)***

Central Precocious Puberty (CPP)

☐ *MKRN3* gene full mutation analysis (38.1) (*Non-GESY*)

☐ *MKRN3* gene known mutation analysis (38.2) (*Non-GESY*)

GLUCOSE AND INSULIN HOMEOSTASIS (MODY AND OBESITY)

☐ Glucose and Insulin Homeostasis *in silico* panel from WES (20.7) *ISO 15189 ***(GESY)***

***GCK* and *HNF1α* Genes, Maturity Onset Diabetes of the Young (MODY)**

☐ *GCK* gene, MODY 2 full mutation analysis (sequencing) (19.1) (*Non-GESY*)

☐ *GCK* gene, MODY 2 analysis for known mutation (sequencing) (19.2) (*Non-GESY*)

☐ *HNF1α* gene, MODY 3 full mutation analysis (sequencing) (20.1) (*Non-GESY*)

☐ *HNF1α* gene, MODY 3 analysis for known mutation (sequencing) (20.2) (*Non-GESY*)

***MC4R* Gene, Obesity**

☐ *MC4R* gene full sequencing (17.1) ***(GESY)***

☐ Obesity, analysis for known mutation (sequencing) (17.2) ***(GESY)***

THYROID FUNCTION

☐ Thyroid Function *in silico* panel from WES (39.1) *ISO 15189 ***(GESY)***

RET-proto-oncogene, Multiple Endocrine Neoplasia type II *ISO 15189

☐ RET mutation analysis (sequencing Exons 10, 11 13 14, 15 & 16) (11.1) ***(GESY)***

☐ RET analysis for known mutation (sequencing) (11.2) ***(GESY)***

OTHER

☐ DNA extraction/storage (96)