



Department of Molecular Genetics, Function & Therapy

Head: Leonidas Phylactou PhD

Referral Letter

Department Code: 27

Patient details

Name: _____ Surname: _____
D.O.B.: ____/____/____ I.D. No.: _____
Nationality: _____ Gender: Male Female
CING No.: _____ Hospital Card No.: _____
Patient Status: GP PP
Address: _____
City: _____ Code: _____ Country: _____
Phone: Home: _____ Work: _____
Fax: _____
e-mail: _____

Referring clinician's / scientist's details

Name: _____ Surname: _____
Hospital / Clinic: _____
Address: _____
City: _____ Code: _____ Country: _____
Phone: _____ Fax: _____
e-mail: _____
Reason for Referral: _____

Signature: _____ Date: ____/____/____

Sample details (Please tick accordingly)

Date and Time of Sample Collection: _____
Sample: Blood CSF Urine Muscle CVS
Other (please specify): _____
 First Investigation Repetition

Sample Receipt (For Laboratory Internal Use)

Received by: _____ Signature: _____
Sample Receipt Date: ____/____/____
Amount: _____ Comments: _____

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

Cystic Fibrosis (CF):

- CF full mutation analysis (1.1)
- CF analysis for known mutation (1.2)
- CF prenatal diagnosis 1st CVS (1.4)
- CF prenatal diagnosis 2nd CVS (1.5)
- CF DNA extraction/storage (1.6)

Familial Mediterranean Fever (FMF):

- FMF full mutation analysis (7.1)
- FMF analysis for known mutation (7.2)
- FMF DNA extraction/storage (7.6)

RET-*proto-oncogene*:

- RET full mutation analysis (11.1)
- RET analysis for known mutation (11.2)
- RET DNA extraction/storage (11.6)

Inherited Deafness:

- Connexin 26 full mutation analysis (sequencing) (13.1)
- Connexin 26 analysis for known mutation (13.2)
- Connexin DNA extraction/storage (13.6)
- Connexin 30 full mutation analysis (sequencing) (15.1)
- Connexin 30 analysis for known mutation (15.2)

Congenital Adrenal Hyperplasia (CAH):

- Full mutation analysis (sequencing) (16.1)
- Analysis for known mutation (16.2)
- CAH DNA extraction/storage (16.6)
- Sequencing per test (exon) (16.7)

Haemochromatosis:

- Haemochromatosis analysis for individual mutations (10.7)

Sweat Test:

- Sweat test (8.7)

Multiple Sclerosis:

- Oligoclonal Bands detection for MS patients (14)

Obesity:

- MC4R full sequencing (17.1)
- Obesity analysis for known mutation (17.2)
- Obesity DNA extraction/storage (17.6)

Focal Segmental Glomerular Sclerosis:

- FSGS family linkage analysis (12.3)

Autosomal Dominant Polycystic Kidney Disease (ADPKD):

- ADPKD analysis for known mutation (2.2)
- ADPKD family linkage analysis (2.3)
- ADPKD prenatal diagnosis 1st CVS (2.4)
- ADPKD prenatal diagnosis 2nd CVS (2.5)
- ADPKD DNA extraction/storage (2.6)

Autosomal Dominant Medullary Cystic Kidney Disease (ADMCKD):

- ADMCKD analysis for known mutation (4.2)
- ADMCKD family linkage analysis (4.3)
- ADMCKD prenatal diagnosis 1st CVS (4.4)
- ADMCKD prenatal diagnosis 2nd CVS (4.5)
- ADMCKD DNA extraction/storage (4.6)

Nephronophthisis:

- Nephronophthisis DNA extraction/storage (5.6)

Cystinuria:

- Cystinuria full mutation analysis (6.1)
- Cystinuria analysis for known mutation (6.2)
- Cystinuria family linkage analysis (6.3)
- Cystinuria prenatal diagnosis 1st CVS (6.4)
- Cystinuria prenatal diagnosis 2nd CVS (6.5)
- Cystinuria DNA extraction/storage (6.6)

Maturity Onset Diabetes of the Young (MODY):

- MODY 2 full mutation analysis (19.1) (Private)
- MODY 2 analysis for known mutation (19.2) (Private)
- MODY3 full mutation analysis (20.1) (Private)
- MODY 3 analysis for known mutation (20.2) (Private)
- MODY DNA extraction/storage (20.6) (Private)

5-alpha Reductase Deficiency (SRD5a):

- SRD5A2 full mutation analysis (21.1) (Private)
- SRD5A2 analysis for known mutation (21.2) (Private)
- SRD5A2 DNA extraction/storage (21.6) (Private)

Growth Hormone Deficiency:

- PROP1 full mutation analysis (22.1) (Private)
- PROP1 DNA extraction/storage (22.6) (Private)

- Other DNA extraction/storage (96)