

Molecular Genetic Testing for Endocrine Disorders by NGS

Please send EDTA blood (min. 4ml adults; 2-4ml children; 1ml neonates) or DNA extracted from Blood EDTA to:
East of Scotland Regional Genetics Laboratory, Level 6 Ninewells Hospital, Dundee DD1 9SY

Clinical Leads: Dr Paul Newey (Endocrinology paul.newey@nhs.scot) & Dr David Goudie (Genetics david.goudie@nhs.scot)

Consultant Clinical Scientist: Dr David Baty (david.baty@nhs.scot)

Lab enquiries: 01382 740533 Tay.esrg@nhs.scot (website www.esrg.scot.nhs.uk)

Please complete all fields as fully as possible and **tick** boxes where appropriate.

Patient Details:

SURNAME:	REQUESTING CONSULTANT:
FORENAME:	TELEPHONE:
D.O.B. / CHI NUMBER	NHS E-MAIL ADDRESS (Reports are sent to NHS accounts as default)
PATIENT'S POSTCODE:	REPORT ADDRESS:
GENDER & ETHNIC ORIGIN:	

Consent: In submitting this sample, the clinician confirms that CONSENT has been obtained: (a) for testing and storage, (b) for the use of this sample and the information generated from it to be shared with members of the patient's family and their healthcare professionals (if appropriate). I confirm I have obtained consent for testing, storage and sharing of information:

.....
Clinician name

.....
Clinician signature

*It is recommended that a 'Record of Discussion' or suitable alternative is completed.

Clinical Details: Please also provide the additional panel-specific clinical information over the page

CLINICAL DIAGNOSIS:	AGE AT DIAGNOSIS:
CLINICAL HISTORY:	
AFFECTED FIRST OR SECOND DEGREE RELATIVE(S): YES <input type="checkbox"/> NO <input type="checkbox"/>	
DETAILS OF RELEVANT FAMILY HISTORY:	
If a variant is known in the family please provide details of the variant and the name and DOB of the affected family member. Please provide a family tree if appropriate	

Endocrine Panels: Please select only one panel. Turnaround time is 56 days unless indicated.

- Hyperparathyroidism/Familial Hypocalcaemic Hypercalcaemia (FHH):
AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET (relevant exons)
- Hypoparathyroidism/Autosomal Dominant Hypocalcaemia (ADH):
AIRE, CASR, GATA3, GCM2, GNA11, PTH, TBCE
- Multiple Endocrine Neoplasia Type 1/4 (MEN1/MEN4)/Familial Isolated Pituitary Adenoma (FIPA):
AIP, CDKN1B, MEN1
- Pheochromocytoma/Paraganglioma (turnaround time 112 days):
FH, MAX, NF1, RET* (relevant exons), *SDHA, SDHB, SDHC, SDHD, SDHAF2, TMEM127, VHL*.
*Features to suggest NF1? YES / NO (If 'NO' analysis will not include *NF1*)

Single gene tests: Please select only one test. Turnaround time is 56 days unless indicated.

- Hyperparathyroidism-Jaw Tumour Syndrome/Parathyroid Carcinoma: *CDC73*
- Multiple Endocrine Neoplasia Type 2 (MEN2A/MEN2B) / Medullary Thyroid Cancer (MTC):
RET exons 5, 8, 10, 11, 13 to 16
- Von Hippel Lindau: *VHL*
- Carney Complex: *PRKAR1A*
- Thyroid Hormone Resistance: *THRB*
- Albright's Hereditary Osteodystrophy/PseudoHypoPara/PseudoPseudoHypoPara: *GNAS*

Phenotype/Clinical Information: Please provide the relevant clinical information for the requested panel

Hyperparathyroidism/Familial Hypocalciuric Hypercalcaemia (FHH) Panel;

c.Ca = PTH =	25OH vit. D =	24hr U. Ca = or U.CCR = (provide local FHH cut off):
Condition suspected: FHH HPTH		Previous HPTH Surgery: Yes / No
Pathology: Adenoma / Hyperplasia Single gland / Multiple glands		

Hypoparathyroidism/Autosomal Dominant Hypocalcaemia (ADH) Panel

Baseline Biochem: cCa ²⁺ = Mg ²⁺ = 25OH-VitD= PTH = PO4 ⁻ =	Condition suspected?
Any associated clinical features?	

Multiple Endocrine Neoplasia Type 1/4 / Familial Isolated Pituitary Adenoma (FIPA) Panel

HPTH: Yes / No Details:	Gastro/Pancreatic - NET: Yes / No Details:
Pit. Tumour: Yes / No Details:	Other relevant tumour type(s): Yes / No Details:

Multiple Endocrine Neoplasia 2A/2B / Medullary Thyroid Cancer (MTC)

MTC: Yes / No Details:	HPTH: Yes / No Details:
Phaeo: Yes / No: Details:	Features of MEN2B: Yes / No Details:

Phaeochromocytoma/Paraganglioma (PPGL):

Features of clinical syndrome?: Yes / No / Not known If Yes: NF1 VHL MEN2A/B other: Details:
Location of PPGL: Adrenal / Extra-adrenal If Adrenal: Unilateral / Bilateral If Extra-Adrenal: Single / Multiple If Extra-Adrenal: Head & Neck Sympathetic chain/intrabdominal Other: Details: Evidence of malignancy? Yes / No / Not known
Family history of PPGL, RCC or GIST; Yes / No Details: