

Hyponadotropic Hypogonadism

Referral Form for Diagnostic Genetics

West of Scotland Genetic Services, Level 2B, Laboratory Medicine, Queen Elizabeth University Hospital, Govan Road, Glasgow, G51 4TF Tel:+44 (141) 354 9330



This form should be completed prior to testing. Please send 5ml of EDTA blood (1ml for neonates) or a DNA specimen (5ug) along with a completed genetic test request form to the address above or email to gg-uhb.geneticdsd@nhs.net If possible, please also store samples locally from the patient's parents to aid variant interpretation.

Results and advice are reported taking into account complex genetic and biochemical information. The interpretation of the results before they are reported depends on the phenotypic data that are provided. This form is therefore best completed by the clinician managing the patient. Clinical letters and laboratory reports, if available, can also aid data interpretation.

Please send completed form to: gg-uhb.geneticdsd@nhs.net

For laboratory advice, please contact the West of Scotland Molecular Genetics Laboratory

Email: geneticlabs@ggc.scot.nhs.uk Tel. 0141 354 9330

Clinical advice: Professor Faisal Ahmed: Faisal.Ahmed@ggc.scot.nhs.uk or Dr Ruth McGowan: ruthmcgowan@nhs.net

Patient Details Forename: _____ Surname: _____ DOB: _____

CHI number/local ID: _____

Referrer Details Lead Clinician: _____ Email: _____

Hospital: _____ City and Country: _____ Telephone: _____ Fax: _____

Address for report: _____ Address for invoice (Non-Scottish Referrals): _____

Suspected Diagnosis: _____

Clinical History of Hypogonadism

Delayed puberty	Pubertal arrest	Infertility
Micropenis	Undescended testes	Hypospadias
Primary amenorrhoea	Early menopause/ovarian failure	Other

History of previous genital surgery (hypospadias, orchidopexy, etc)

Details:

History of coexisting pituitary hormone deficiency:

GH	TSH	Prolactin	ACTH	ADH
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Family History

Consanguinity: _____ Anosmia _____ Delayed Puberty _____ Infertility _____ DSD _____ Other _____

Details:

Parental sample collected: _____

Physical findings**Date**

Weight: (kg)

Height: (cm)

Micropenis:

Stretch penile length (cm):

Urethral opening:

Labioscrotal fusion:

Undescended testes:

Position of testis (Right)

(Left)

Testicular volume (ml):

Gynaecomastia:

Genital Tanner staging:

Pubic hair tanner staging:

Breast Tanner staging:

Sense of smell:

Microcephaly:

Coloboma:

Cleft lip/palate:

Dental Agensis:

Synkinesia:

Ataxia:

Skeletal anomalies:

Hearing impairment:

Pigmentation abnormalities:

Neurodegenerative disorder:

Renal agenesis:

Other details:

Random and stimulated hormone measurements

Date				Date	
LH (IU/l)				Peak LH (IU/l) (LHRH test)	
FSH (IU/l)				Peak FSH (IU/l) (LHRH test)	
Testosterone (nmol/L)				Peak Testosterone (nmol/l) (hCG test)	
Cortisol (nmol/l)				Peak cortisol (nmol/l) (synacthen)	
IGF-1 (nmol/l)				Peak GH (µg/l) (GH stimulation)	
Estradiol (pmol/l)				FT4 (nmol/l)	
AMH (pmol/l)				TSH (mU/l)	
ACTH (pmol/l)				Prolactin (ug/l)	
Other:					

MRI findings

Pituitary size:

Bright spot:

Pituitary stalk:

Olfactory nerve/sulcus:

Other CNS abnormalities:

Previous genetic results

Karyotype:

Array:

DNA stored:

Previous analysis of HH genes: Y/N

Result:

Other genetic analysis:

Date of form completion:

Name:

Save form**Print form****Email**

DSD Diagnostic Service – internal use only. Please leave this blank

Date	Discussion	Initials

Version 1

Issue date 02/04/20

Review date 02/04/22