Hypogonadotropic 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

NHS Greater Glasgow and Clyde

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 molgen@ggc.scot.nhs.uk If possible, please also store samples locally from the patient's parents to aid variant interpretation. First line investigation for sex chromosome abnormality is targeted chromosome analysis (TCA). If not already complete, please send 3ml of blood in Lithium Heparin to your local laboratory for analysis.

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 forms to molgen.genetic@nhs.scot
For laboratory advice, please contact the West of Scotland Molecular Genetics Laboratory. Email: molgen.genetic@nhs.scot Tel: 0141 354 9330
Clinical advice: Professor Faisal Ahmed: Faisal.Ahmed@nhs.scot or Dr Ruth McGowan: Ruth.McGowan@nhs.scot

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

Family History

Consanguinity: Please select ☐ Anosmia ☐ Delayed puberty ☐ Infertility ☑ DSD ☑ Other

Details:

Parental sample collected: Please select

Physical findings **Date**

Weight: (kg) Height: (cm)

Micropenis: Please select Stretch penile length (cm): Urethral opening: Please select

Labioscrotal fusion: Please select Undescended testes: Please select Position of testis (Right) Select (Left) Select

Testicular volume (ml): Gynaecomastia: Please select Genital Tanner staging: Please select

Pubic hair tanner staging: Please select Breast Tanner staging: Please select

Sense of smell: Please select Microcephaly: Please select Coloboma: Please select

Cleft lip/palate: Please select Dental Agenesis: Please select Synkinesia: Please select

Ataxia: Please select Skeletal anomalies: Please select Hearing impairment: Please select

Pigmentation abnormalities: Select Neurodegenerative disorder: Select Renal agenesis: Please select

Other details:

Random and stimulated hormone measurements

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

MRI findings Pituitary size: Please select Bright spot: Please select Pituitary stalk: Please select

Olfactory nerve/sulcus: Please select Other CNS abnormalities:

Previous genetic results Karyotype: CGH: DNA stored: Please select

Previous analysis of HH genes: Y/N Please select 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