Referral Form for DSD Diagnostic Service

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 **(http://www.nhsggc.org.uk/media/236026/geneticstestrequestonlineform-pdf.pdf)** to the address above. For panel testing, please also send samples 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 avialable, 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):

Provisional Diagnosis

Birth weight: Birth gestation: Sex assignment: Karyotype:

Suspected diagnosis:

Associated conditions

Family History Family history of DSD: -Please select- Other family history: -Please select-

Parental consanguinity: -Please select-

Any other information:

Clinical Features on External Examination Date of examination:

Labioscrotal fusion: -please select- Urethral opening: -please select- Phallus: -please select-

Stretch Length (mm): Position of gonads: Left: -please select- Right: -please select-

Gynaecomastia: -please select- Any other information:

Clinical Features on Internal Examination Date of examination:

Uterus present: Normal Fallopian tube (left): -please select- Fallopian tube (right): -please select-

Urogenital Sinus: None Vas Deferens (left): -please select- Vas Deferens (right): -please select-

Any other information:

Description of gonads

	Normal testes	Normal Ovary	Ovotestis	Dysplastic testis	Streak	Gonads absent
Left:	☐	☐	☐	☐	☐	☐
Right:	☐	☐	☐	☐	☐	☐

Any other information:

Biochemistry Date of birth:

Random/Spot measurements:

Date				
AMH pmol/l				
Inhibin B ng/l				
Testosterone nmol/l				
Oestradiol pmol/l				
Andro'dione nmol/l				
17OHP nmol/l				
DHAS umol/l				
DHT nmol/l				
LH iU/l				
FSH iU/l				

HCG Stimulation Test: If other please state:

Date			
Testosterone nmol/l			
Androstenedione nmol/l			

LHRH stimulation test:

Date			
Minutes	0	20-30	60
LH iU/l			
FSH iU/l			

Adrenal Stimulation Test:

Date			
Minutes	0	20-30	60
Cortisol nmol/l			
17 OHP nmol/l			

Urine steroid Profile: -please select- Provide further details:

Results:

QF-PCR: -please select-

Karyotype: -please select-

Microarray: -please select-

DNA stored:

Other genetic analysis: -please select-

Parental samples:

Father Forename: Surname: DOB:

Mother Forename: Surname: DOB:

Relevant clinical information

Date of form completion: Name:

Save form Print form email form

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

Date	Discussion	Initials