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

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.

Results and advice are reported taking into account complex genetic and biochemical information. It is therefore important that we capture as much clinical information regarding the DSD phenotype as possible. This form is therefore best completed by the clinician managing the DSD. Clinical letters and laboratory reports, if available, can also aid data interpretation.

Please send completed form to: geneticlabs@ggc.scot.nhs.uk
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@ggc.scot.nhs.uk

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:

Cl·Yſfê Iſăű2Mè 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-

Stretched 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: -please select- Fallopian tube (left): -please select- Fallopian tube (right): -please select-

Urogenital Sinus: -please select- Vas Deferens (left): -please select- Vas Deferns (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				
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			
Andro'dione 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