

CORE DATASET – ESPE DSD REGISTRY

Revision date								
Revision author								
Year of Birth								
Local Hospital Identification				Date of First Notification				

Centre Reporting Clinician

Clinical Presentation

Sex assigned Male ☐ Female ☐ NK ☐

Phallus Size Normal Male ☐ Small Male ☐ Large Male ☐
Normal Female ☐ Large Female ☐ Chordee ☐ NK ☐

Phallus Length Date..... Lengthmm NK ☐

Urinary Meatus Normal Male ☐ Hypospadias ☐ Distal ☐ Mid ☐ Proximal ☐
Normal Female ☐ NK ☐ (glanular, coronal) (penile shaft) (penoscrotal, scrotal, perineal)

Labioscrotal Fusion Yes ☐ No ☐ NK ☐

Right Gonad Labioscrotal ☐ Inguinal ☐ Abdominal ☐ Absent ☐ Undescended-Nonspecific ☐ NK ☐
Left Gonad ☐ ☐ ☐ ☐ ☐ ☐

External Masculinisation Score ____ (0 – 12) NK ☐ **Tanner Stage** ____ (1 - 5) NK ☐

Internal Sex Organs Mullerian <input type="checkbox"/> Wolffian <input type="checkbox"/> NK <input type="checkbox"/>	Modality US <input type="checkbox"/> MRI <input type="checkbox"/> CT <input type="checkbox"/> LS <input type="checkbox"/> LT <input type="checkbox"/>
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Associated Malformation Adrenal ☐ GIT ☐ Renal ☐ Haematological ☐ Skeletal ☐ CNS ☐ ENT ☐ Resp ☐
Heart ☐ SGA ☐ Short stature ☐ Craniofacial ☐ Non-defined Syndrome ☐
Other ☐

DSD Classification

Karyotype 46XY ☐ 46XX ☐ Presumed XY ☐ Presumed XX ☐
46XX/46XY ☐ 45X ☐ 45X/46XY ☐ Other ☐

Disorder Type Disorder of Gonadal Development ☐ Defect of Mullerian Development ☐
Disorder of Androgen Synthesis ☐ Persistent Mullerian Duct Synd ☐
Disorder of Androgen Action ☐ Leydig Cell Defect ☐
Disorder of Androgen Excess ☐ Cloacal Dystrophy ☐
Nonspecific Undermasculinisation Disorder ☐ Other ☐

Actual Diagnosis

Certainty of Diagnosis Based On

Clinical Features	Certain <input type="checkbox"/>	Uncertain <input type="checkbox"/>	
Biochemistry	Certain <input type="checkbox"/>	Uncertain <input type="checkbox"/>	NA <input type="checkbox"/>
DNA Analysis	Certain <input type="checkbox"/>	Uncertain <input type="checkbox"/>	NA <input type="checkbox"/>

DNA Analysis

Performed	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Abnormality Found	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/> NA <input type="checkbox"/>
Further studies	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/> NA <input type="checkbox"/>

Availability of Further Information

Case Notes	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Growth Data	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Puberty Data	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Urine	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Serum	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
DNA	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Tissue	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Cell line	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>

Family

Parental Consanguinity	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
History of DSD	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
History of Infertility	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>
Availability of Samples	Yes <input type="checkbox"/>	No <input type="checkbox"/>	NK <input type="checkbox"/>

Free Text

Register ID							
Received							
Entered							
Info Sheet 1							
Info Sheet 2							
Removed							

Future Contact – No ☐

Please retain one copy in case notes and send another copy to:-

Further copies & Guidance available from Register Coordinator

Please file all revision copies together in case notes

V.5.03.08