

## Guidance for Completing the EuroDSD Register Core Dataset



### About EuroDsD

The EuroDSD project is a 3-year EU funded project funded under the FW7 ICT e-Health programme and will improve our understanding of disorders of sex development (DSD). These conditions are relatively rare and require pooling of as much clinical and genetic information as possible. The National e-Science Centre at the University of Glasgow is developing and supporting an e-Infrastructure through which secure access to a range of genotypic and phenotypic data across partner sites across Europe will be made available. This core data set and the registry through which it will be made available through secure upload and shared with authorized individuals provides the starting point for sharing of clinical information and improving clinical care.

### DSD Identifiers

#### Centre

Choose or add details of the base of the reporting consultant.

#### Local ID

This is the ID of the record, usually maintained by local electronic hospital systems.

#### Date of First Presentation

Date of first notification of case to Register in Day (- -) / Month (- -) / Year (- -).

#### Clinician

Surname of reporting consultant

#### Contact

Contact person for more information of this patient.

### Clinical Presentation

#### Last Revision Date & Author

It is possible that case details may change after first notification. This is the date when a revised dataset are sent to the hospital. The author of the revised report needs to enter surname.

#### Phallus Length

Enter length of phallus in millimetres and the date of measurement. Select 'MK' if phallus length not available.

#### Phallus Size

Select based on general appearance of phallus. Please also select box if Chordee present. Select 'NK' if there is no information available on phallus size.

#### Urinary Meatus

Select 'Normal Male' or 'Normal Female' if urinary meatus is normally sited for a boy or a girl, respectively.

If Urinary meatus is abnormally sited on a phallus, select 'hypospadias' AND select type of hypospadias. Select 'hypospadias' and 'NK' if type of hypospadias is unclear. Select 'NK' if there is no information on urinary meatus.

#### Labioscrotal Fusion

Select based on whether there is labioscrotal fusion or not. Select 'NK' if this information is not available.

#### Gonads

Select appropriate position of gonads. Select 'labioscrotal' for gonads that are retractile or clearly present anywhere within the labioscrotal folds. Gonads that are above the scrotal sacs and in the inguinal region are classified as 'inguinal'. Select 'Abdominal' for gonads that are impalpable but identified to be present internally. Select 'Absent' if gonads are impalpable and cannot be identified. Select 'Undescended-Nonspecific' if the gonads have not descended but their position is unclear. Select 'NK' if there is no information on position of gonads.

### Internal Sex Organs

#### Mullerian & Wolffian Structure

Specify whether the patient has Mullerian (Uterus, Fallopian Tubes, Upper part of vagina) and/or Wolffian (vas deferens, seminal vesicles and the ejaculatory ducts) structures. Select both boxes if both structures are present. Please select 'NK' if this information is not available.

#### Modality

Select modalities that have been used to identify internal sex organs – US scan, MRI scan, CT Scan, LS – Laparoscopy, LT – Laparotomy.

#### External Masculinisation Score (EMS)

The EMS is an objective method of scoring the extent of masculinisation of the external genitalia. The score for normal external male genitalia is 12. It is based on phallus size, location of urinary meatus, labioscrotal fusion and location of gonads. The score for each of these is in parentheses in the table below. The EMS cannot be calculated if information on all these features is not available.

Phallus	Urinary Meatus	Labioscrotal Fusion	Right Gonad	Left Gonad
Normal for male (3)	Normal for male (3)	Yes (3)	Labioscrotal (1.5)	Labioscrotal (1.5)
Small for male (0)	Hypospadias - Distal <sup>a</sup> (2)	No (0)	Inguinal (1)	Inguinal (1)
Large for male (3)	Hypospadias - Mid <sup>b</sup> (1)	Not Known	Abdominal (0.5)	Abdominal (0.5)
Normal for female (0)	Hypospadias - Proximal <sup>c</sup> (0)		Absent (0)	Absent (0)
Large for female (3)	Normal for female (0)		Non-specific undescended	Non-specific undescended
Not Known	Not Known		Not Known	Not Known

<sup>a</sup> Distal – Glanular, Coronal; <sup>b</sup> Mid – Midshaft, Penile; <sup>c</sup> Proximal – Penoscrotal, Scrotal, Perineal

#### Tanner Stage

Available options include 1, 2, 3, 4, and 5.

### Disorder of Sex Development (DSD) Classification

#### Sex Assigned

This is the sex of rearing. Please select male for boy, female for girl or not known.

#### Karyotype – Primary Root

This is the primary root of the DSD Classification and is based on the peripheral karyotype from a blood sample. If there is a karyotype from other tissues please provide further information in Free Text. Examples may include 46XX, 46 XY, 45X, 47XXY, 45X/46XY, 46XX/46XY. 'Presumed 46XY' may be selected in those cases of undermasculinised boys where the clinician suspects that clinically the boy should not have an abnormality of his karyotype.

#### Disorder Type – Second Root

This is the secondary root of the DSD Classification and is generally based on the broad type of anticipated disorder. This part of the classification system will guide the user into specific lists of actual diagnoses within each disorder type.

### Actual Diagnosis

Disorder of gonadal development	Disorder of androgen synthesis	Disorder androgen action	Disorder of Androgen excess	Leydig cell defect	Persistent Mullerian Duct Syndrome	Defects of Mullerian development	Non-specific Undermasculinisation Disorder	Other
Complete gonadal dysgenesis	StAR def	PAIS	21 $\alpha$ hydroxylase def (CYP21A)	Leydig cell hypoplasia	AMH low	MURCS	Isolated hypospadias	Cloacal Anomaly
Partial gonadal dysgenesis	P450 scc def (CYP11A1)	CAIS	11 $\beta$ hydroxylase def (CYP11B1)	LH deficiency	AMH normal	MRKH	Isolated bilateral cryptorchidism	Bladder Exstrophy
Gonadal regression	3 $\beta$ -HSD def (HSD3B2)	Other	Aromatase def (CYP19A1)	Other	AMH not known	Uterine Didelhy's Other	Combined anomalies EMS > 8	Smith Lemli Opitz Synd
Ovotesticular DSD	CYP17 def (P450CYP17)		P450 oxidoreductase def (POR)				Combined Anomalies EMS 5-8	Other
Testicular DSD	17 $\beta$ HSD def (HSD17B3)		Maternal androgens				Combined Anomalies EMS < 5	
Other	5 $\alpha$ reductase def (SRD5A2)		Other					
	P450 oxidoreductase def (POR)							
	Other							

### Associated Malformations

Please tick box if there are any confirmed or suspected abnormality in another system. Available options are:

CNS – Central Nervous System

Heart

Renal

Digestive

Skeletal

Skin

ENT – Ears, Nose, Throat

Blood and Lymph

Craniofacial

Adrenal

GIT – Gastrointestinal Tract

Haematological

Respectfully yours

SGA – Small for Gestational Age

Short Stature

Non-Definded Syndrome

Other

### Certainty of Diagnosis

Specify the level of certainty of diagnosis based clinical, biochemical and molecular genetic findings

### Genetic Certainty

Available options are Certain and Uncertain

### Clinical Features

Available options are Certain and Uncertain

**Biochemistry**

Available options are Certain, Uncertain, and Not Applicable

**DNA Analysis**

Available options are Certain, Uncertain, and Not Applicable

**DNA Analysis**

Specify whether any DNA analysis has been performed and the outcome. Details of genes analysed and results can be included in Free Text. Further studies such as 'studies of gene function', comparative genome hybridisation' can be also mentioned here.

**DNA Analysis Performed**

Available options are Yes, No, and Not Known

**Abnormality Found**

Available options are Yes, No, Not Known, and Not Applicable

**Further Studies**

Available options are Yes, No, Not Known, and Not Applicable

**Material - Availability of Further Information**

Indicate whether following information or samples are available. Case Notes, Growth Data, Puberty Data, Urine, Serum, DNA, Tissue, Cell line. Specify origin (gonadal, skin, genital skin) of tissue or cell line in Free Text.

**Clinical Information**

Available options are Yes and No

**Case Notes**

Available options are Yes, No, and Not Known

**Growth Data**

Available options are Yes, No, and Not Known

**Puberty Data**

Available options are Yes, No, and Not Known

**DNA**

Available options are Yes, No, and Not Known

**Tissue**

Available options are Yes, No, and Not Known

**Cell Line**

Available options are Yes, No, and Not Known

**Urine**

Available options are Yes, No, and Not Known

**Serum**

Available options are Yes, No, and Not Known

**Family**

Indicate whether there is any known family history of DSD or infertility and whether there are any samples for analysis on any family members. If so, provide further details in Free Text.

**Parental Consanguinity**

Available options are Yes, No, and Not Known

**History of DSD**

Available options are Yes, No, and Not Known

**History of Infertility**

Available options are Yes, No, and Not Known

**Availability of Samples**

Available options are Yes, No, and Not Known

**Free Text**

Add any other information here briefly. These data may be difficult to search.

**Section for Auditing Purposes**

This section is only for system monitoring and auditing.

**Removed**

For cases who do not want to be included on the Register, this is the date when the data were removed from the Register and returned to the Reporting Clinician

**No Future Contact**

Patient may choose whether needing future contacts. Available options are Yes and No.

**Uploader**

Person who uploads the data

**Upload Time**

Date when the data were entered into the Register in format: Day (- -) / Month (- -) / Year (- -) Hour(- -) : Minute (- -)

**Range of Sharing of this record**

To specify the range of sharing of this record. Available options include country, EU, and everybody.

The following 2 items might be limited to Scotland hospital systems.

**Info Sheet 1**

Date when the first Register information sheet is sent out to the case and parents if case less than 16 yr old

**Info Sheet 2**

Date when the Register information sheet should be sent out to a case who was less than 16 yrs old at initial registration.

### Revising the information

Information on the EuroDSD Register can be revised as necessary. If any data is revised, the date of revision and the surname of the author should be modified accordingly.

### Abbreviations

AMH	Anti-Mullerian Hormone
CAIS	Complete Androgen Insensitivity Syndrome
CHI	Community Health Index
CNS	Central Nervous System
CT	Computed Tomography
ENT	Ears, Nose, Throat
GIT	Gastrointestinal Tract
LS	Laparoscopy
LT	Laparotomy
MRI	Magnetic Resonance Imaging
MRKH	Mayer Rokitansky Kuster Hauser Syndrome
MURCS	Mullerian hypoplasia/aplasia, renal agenesis and cervicothoracic somite dysplasia
NA	Not available
NK	Not Known
PAIS	Partial Androgen Insensitivity Syndrome
SGA	Small for Gestational Age
US	Ultrasound