Guidance for Completing the I-DSD Register Core Dataset

About I-DSD

The I-DSD Registry was developed initially through the EuroDSD project with funding from the EC FP7 and is currently supported by a 5 year grant from MRC. The Registry was established in order to 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 around the world 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 miliimetres 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.

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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 [a] (2)	No (0)	Inguinal (1)	Inguinal (1)
Large for male (3)	Hypospadias - Mid [b] (1)	Not Known	Abdominal (0.5)	Abdominal (0.5)
Normal for female (0)	Hypospadias - Proximal [c] (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

[a] Distal – Glanular, Coronal; [b] Mid – Midshaft, Penile; [c] 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.

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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	21αhydroxylase def (CYP21A)	Leydig cell hypoplasia	AMH low	MURCS	Isolated hypospadias	Cloacal Anomaly
Partial gonadal dysgenesis	P450 scc def (CYP11A1)	CAIS	11βhydroxylase def (CYP11B1)	LH deficiency	AMH normal	MRKH	Isolated bilateral cryptorchidism	Bladder Exstrophy
Gonadal regression	3β-HSD def (HSD3B2)	Other	Aromatase def (CYP19A1)	Other	AMH not known	Uterine Didelhys	Combined anomalies EMS > 8	Smith Lemli Opitz Synd
Ovotesticular DSD	CYP17 def (P450CYP17)		P450 oxidoreductase def (POR)			Other	Combined Anomalies EMS 5-8	Other
Testicular DSD	17βHSD def (HSD17B3)		Maternal androgens				Combined Anomalies EMS < 5	
Other	5α 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

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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.

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Revising the information

Information on the I-DSD 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	Laparatomy
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