

I-DSD Registry & I-CAH Registry Update

faisal.ahmed@glasgow.ac.uk

www.i-dsd.org

www.i-cah.org



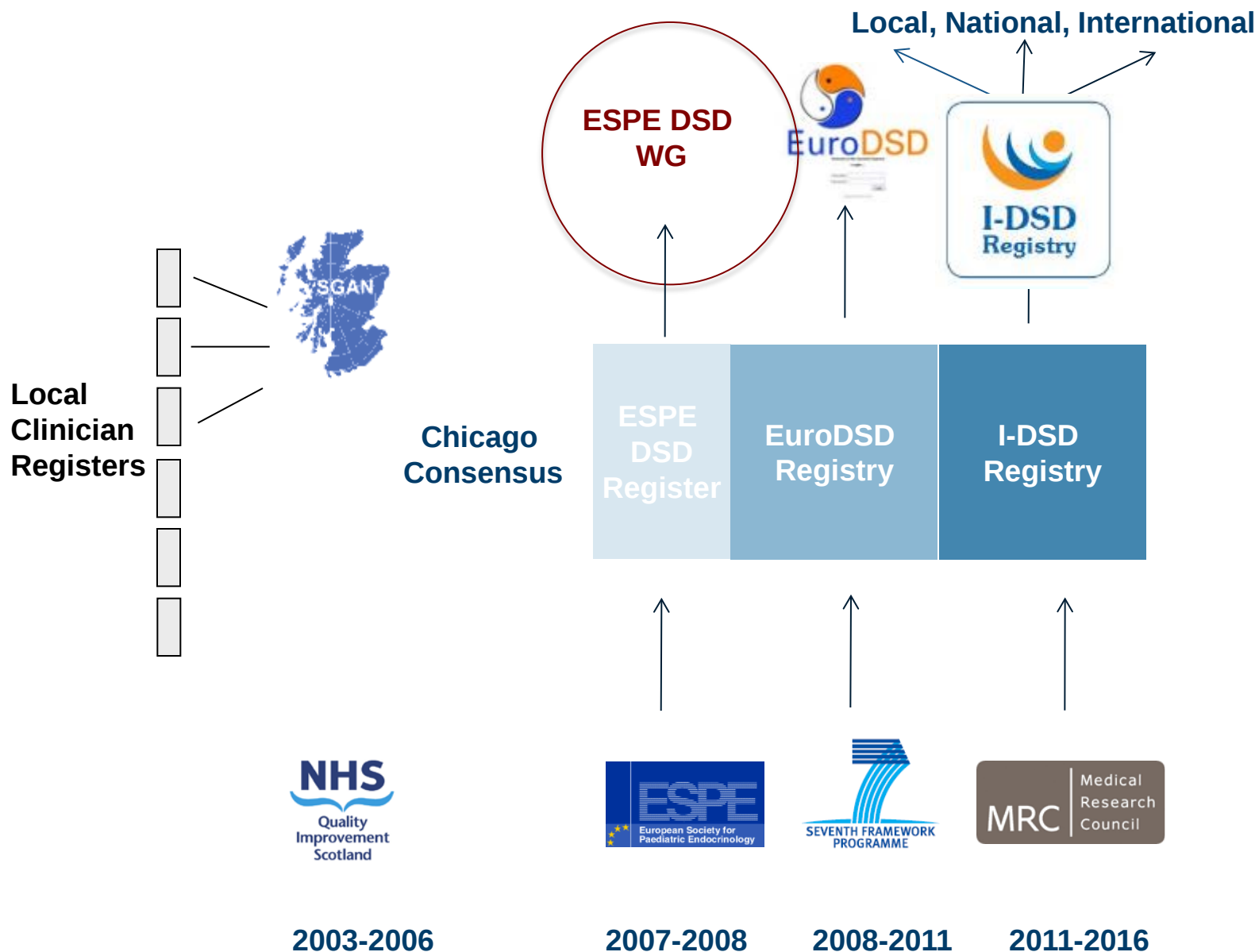
- **Collection of standardised clinical information on rare conditions:**
 - Understand pathogenesis
 - Improve diagnostic yield
 - Understand short-term and long-term outcome
 - Assess quality of care
 - Plan and perform clinical trials
- **Facilitate and assess collaboration through a network of centres**
- **One of the core criteria for EU Centres of Expertise in Rare Diseases (EUCERD)**

Mission, Vision and Scope of RD ERNs

- ERN to cover core tools and activities
 - Registries
 - quality assurance mechanisms for laboratory testing
 - mechanism for information flow for good practice guidelines
 - training/education tools
 - mechanisms for evaluation/indicators of performance
 - communications infrastructure
 - cross-border referral mechanisms
 - telemedicine core

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I-DSD Users

- Members of professional medical/scientific societies

I-DSD Clinical Centre Leads

- Responsible for data entry – governance and quality control
- View, modify, delete all data
- View core data on all cases – (future – fee)

I-DSD Research Users

- Approval by PMG after completion of application form
- Steering committee involvement if overlap with another project
- 6 months access – renewable on submission of progress report
- (Future – fee per module access, project groups for modules)

I-DSD Patient/Parent Participants

- Access following registration of case
- View own core data
- Future –personalised sign-posting & patient involvement

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[Research Results](#)
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Search registered users

Full Name:

Country & Centre:

--- Please Select ---

Email:

Position:

Primary Role:

--- Select Your Primary Role ---

- ☐ Biochemical Evaluation
- ☐ Genetic Evaluation
- ☐ Clinical Evaluation
- ☐ Medical Care
- ☐ Surgical Care
- ☐ Psychological Care
- ☐ Genetic Counselling

Secondary Roles:

Professional Society membership:

- ☐ Gonadal Development
- ☐ Androgen Synthesis
- ☐ Androgen Action
- ☐ Androgen Excess
- ☐ Mullerian Development

Areas of Interest:

Algeria	Iran
Argentina	Israel
Australia	Italy
Austria	Jordan
Belgium	Kazakhstan
Bosnia & Herzegovina	Kuwait
Brazil	Lithuania
Bulgaria	Malawi
Canada	Malaysia
China	Mexico
Colombia	Morocco
Cyprus	Netherlands
Czech Republic	Nigeria
Denmark	Pakistan
Egypt	Poland
Estonia	Romania
France	Russia
Georgia	Serbia
Germany	Spain
Hong Kong	Sri Lanka
Hungary	Sudan
India	Sweden
Indonesia	Switzerland
	Turkey
	USA
	United Arab Emirates
	United Kingdom

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--- Please Select ---

Email:

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Primary Role:

--- Select Your Primary Role ---

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Argentina 1

Israel 2

Italy 2

Belgium 1

Kuwait 1

Brazil 2

China 1

Morocco 1

Czech Republic 1

Pakistan 1

Poland 2

Egypt 3

Romania 2

France 1

Spain 1

Germany 5

Sudan 1

Sweden 1

Switzerland 4

Turkey 4

United Kingdom 6

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[Home](#)
[Records](#)
[Statistics](#)
[Research Results](#)
[My Account](#)
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There are currently **1881** records stored in the registry.

Centre	Country	Data for local institution access Only	Data For national access Only	Data for EEA access only	Data for access by all international collaborators	Total
Cambridge	United Kingdom	0	162	47	17	226
Istanbul University	Turkey	1	0	63	136	200
Rotterdam	Netherlands	71	0	107	0	178
Luebeck	Germany	11	7	52	48	118
Ghent	Belgium	0	0	112	2	114
Cairo National Research Center	Egypt	4	1	90	5	100
Glasgow	United Kingdom	6	19	6	69	100
Pisa	Italy	13	6	73	5	97
Bologna	Italy	54	0	32	8	94
Stockholm	Sweden	4	10	44	0	58
Magdeburg	Germany	0	0	0	56	56
Sheffield Hallamshire	United Kingdom	1	0	0	49	50
Bern	Switzerland	46	0	0	2	48
Lyon	France	26	0	18	0	44
Nijmegen	Netherlands	30	0	0	14	44
Istanbul Marmara	Turkey	0	0	0	43	43
Prague	Czech Republic	0	1	39	1	41
Birmingham Children's Hospital	United Kingdom	0	0	0	41	41
Berlin	Germany	0	0	0	23	23
Birmingham	United Kingdom	3	0	19	0	22
Khartoum	Sudan	0	0	0	21	21

I-DSD (English)

- [I-DSD Registry Information Leaflet & Consent Form](#)
- [I-DSD Registry Information Leaflet Child](#)
- [I-DSD consent form \(word\)](#)

Argentina

- [CEI-aprob I-DSD](#)
- [Registro I-DSD AI 14-17 años](#)
- [Registro I-DSD AI 7-13 años](#)
- [Registro I-DSD CI Padres y adultos](#)
- [Registro I-DSD SOP versión 2](#)

Belgium

- [IC_ouders](#)
- [IC_patient](#)

Brazil

- [Brazil \(Portuguese\) Information sheet for adults](#)
- [Brazil \(Portuguese\) Information sheet for child](#)
- [Brazil \(Portuguese\) Consent Form \(Word\)](#)
- [Brazil TCLE GIEDDS](#)

China

- [China informed consent \(chinese\)](#)

Egypt

- [Egypt consent form \(arabic\)](#)

France

- [Info Eclairée Patients EuroDSD](#)

Germany

- [Aufklärung_HL](#)
- [Einverständnis_HL](#)

Israel

- [Israel טופס הסכמה ליישום מאגר DSD](#)

Italy

- [informativa e consenso](#)
- [Scheda_registro_cartaceo](#)

Morocco

- [Morroco Copie de consentement park \(arabic\)](#)

Netherlands

- [Informatie_brief_vanaf 16 jaar_I_DSD_I_CAH registry version 1,0](#)
- [Informatie_brief_jongeren 12 tot 16 jaar_I_DSD I_CAH registry version 1,0](#)
- [Informatie_brief_oudersverzorgers_I_DSD_I_CAH registry version 1,0](#)

Poland

- [Poland Informed Consent](#)

Russia

- [Информационный лист о Регистре I-DSD для взрослых](#)
- [Информационный лист о Регистре I-DSD для детей](#)
- [Согласие на ввод данных в Регистр I-DSD](#)

Romania

- [Information sheet for adult](#)
- [Information Sheet for child](#)

Spain

- [CI I-DSD-Diciembre 2014.pdf](#)
- [Spanish Information sheet for adolescents](#)

Switzerland

- [Switzerland 20131121_IC_DSD](#)
- [Switzerland 20131121_StudyInfo_DSD](#)

Turkey

- [Aufklärung_HL Turkish](#)
- [Einverständnis_HL Turkish](#)

I-CAH Registry Patient Information Sheets

[I-CAH Information sheet Adult & Consent form](#)

[I-CAH Information sheet Child](#)

[I-CAH consent form](#)

The Patient Information Sheets have been approved by the West of Scotland Research Ethics Committee.

Word versions available upon request from the Registry Manager, lillian.bryce@glasgow.ac.uk

- Cox K, Bryce J, Jiang J, Rodie M, Sinnott R, Alkhawari M, Arlt W, Audi L, Balsamo A, Bertelloni S, Cools M, Darendeliler F, Drop S, Ellaithi M, Guran T, Hiort O, Holterhus P-M, Hughes I, Krone N, Lisa L, Morel Y, Soder O, Wieacker P, Ahmed SF. **Novel associations in disorders of sex development: findings from the I-DSD Registry.** J Clin Endocrinol Metab. 2014.
- Kolesinska Z, Ahmed SF, Niedziela M, Bryce J, Molinska-Glura M, Rodie M, Jiang J, Sinnott RO, Hughes IA, Darendeliler F, Hiort O, van der Zwan Y, Cools M, Guran T, Holterhus P-M, Bertelloni S, Lisa L, Arlt W, Krone N, Ellaithi M, Balsamo A, Mazen I, Nordenstrom A, Lachlan K, Alkhawari M, Chatelain P, Weintrob N. **Changes over time in sex assignment for disorders of sex development.** Pediatrics. 2014
- Ahmed SF, Bryce J, Hiort O. **Clinical & research networks in DSD, in Differences & Disorders of Sex Development.** Ed Hiort O & Ahmed SF, Karger 2014

5th International DSD Symposium

New Zebra, Ghent, Belgium

11th-13th June 2015



Total Attendees – 220 (multiple disciplines)

Argentina, Australia, Austria, Belgium, Brazil, Bulgaria, Canada, Cyprus, Denmark, Egypt, France, Germany, Indonesia, Israel, Italy, Kazakhstan, Norway, Poland, Romania, Russia, Sweden, Switzerland, Turkey, UK, USA,

Training workshop

Invited Speakers – 24

Oral communications – 32

Poster communications - 48

‘Active Participants’:Attendees – 50%

2011-Luebeck



2013-Glasgow



2015-Ghent



Clinical I-DSD Registry User - £50

Non-clinical user - £35

Non-user - £70

Concessions - £30

Conference Dinner - £35

Best Long Oral Presentation D Baetens, Belgium

Identification and functional characterization of ESR2, a new disease gene for 46,XY DSD

Best Short Oral Presentation A Morris, Australia

CAH in Melbourne: Surgical timing and complications, with outcomes including body image and genital sensation in a cohort study

Best Poster Presentation A Zainuddin, Indonesia

Body image and genitalia appearance concerns and virilization features of CAH female patients in Malaysia.

Runner Up Poster Presentation N de Neve-Enthoven,
Netherlands

Psychosocial well-being in Dutch adults with a DSD

P-1-61

Current Models of Practice & Professional Development of Clinicians in DSD Centres - Results from an International Survey of Specialist Care for DSD

Andreas Kyriakou^a, Arianne B Dessens^b, Jillian Bryce^a, Ira Haraldsen^c, Violeta Iotova^d, Anders Juul^e, Maciej Krawczynski^f, Agneta Nordenskjöld^g, Marta Rozas^h, Caroline Sandersⁱ, Olaf Hiort^j & S Faisal Ahmed^a

P-2-307

Diagnostic Approach to a Newborn with Suspected DSD: Results From an International Survey of Specialist Care for DSD

Andreas Kyriakou^a, Arianne B Dessens^b, Jillian Bryce^a, Ira Haraldsen^c, Violeta Iotova^d, Anders Juul^e, Maciej Krawczynski^f, Agneta Nordenskjöld^g, Marta Rozas^h, Caroline Sandersⁱ, Olaf Hiort^j & S Faisal Ahmed^a

P-1-58

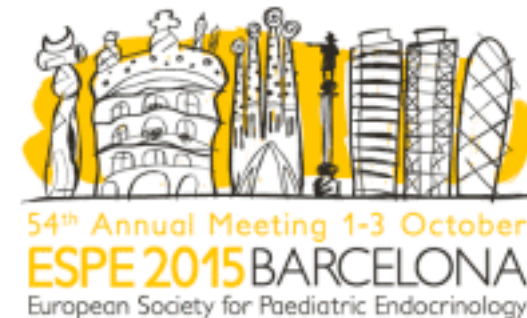
Long-term Endocrine Outcome in Men with Partial Androgen Insensitivity Syndrome

Angela K Lucas-Herald^a, S Faisal Ahmed^a, Silvano Bertelloni^b, Anders Juul^c, Jillian Bryce^a, Jipu Jiang^a, Martina Rodie^a, Marie L Johansen^c, Olaf Hiort^d, Paul-Martin Holterhus^e, Martine Cools^f, An Desloovere^f, Naomi Weintrob^g, Sabine E Hannema^h, Tulay Guranⁱ, Feyzad Darendeliler^j, Anna Nordenstrom^k & Ieuan Hughes^l

P-2-312

Birth Weight in Different Aetiologies of Disorder of Sex Development

Sukran Poyrazoglu^a, Feyza Darendeliler^a, Syed Faisal Ahmed^b, Jillian Bryce^b, Jipu Jiang^b, Martina Rodie^b, Olaf Hiort^c, Sabine E Hannema^d, Silvano Bertelloni^e, Lidka Lisa^f, Tulay Guran^g, Ieuan Hughes^h, Martine Coolsⁱ, Pierre Chatelain^j, Hedi L Claahsen-van der Grinten^k, Anna Nordenstrom^l, Paul-Martin Holterhus^m, Birgit Köhlerⁿ, Marek Niedziela^o & Nils Krone^p



This is the exact view that the participant will see for Record ID 21 should you approve this access

Core Information

Register ID	21
Country & Centre	United Kingdom Glasgow
Year of Birth	2007
Age of First Presentation	< 1 month
Clinician	Ahmed
Contact	faisal.ahmed@glasgow

DSD Classification

Sex Assigned	M
Karyotype	46 XX
Disorder Type & Actual Diagnosis	Disorder of gonadal d Testicular DSD
Birth Weight (Gram)	3200
Associated Conditions	Craniofacial

To grant participant access to this record, please provide the participant's email address:

Grant Participant Access

Cancel

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Scottish Disorders of Sex Development
NMCN



Home News Executive Clinical Area Publications Membership CAS Links

Welcome

Welcome to the website for the Scottish Disorder of Sex Development National Managed Clinical Network.

Within the website you will find:

- Current Network Activities
- Information on events
- Network publications
- Links to relevant organisations
- Patient/Carers web page

If you have anything you would like added or amended on the website or would be interested in helping with content please contact nss.sdsd@nhs.net

NEWS & EVENTS
[Scottish Audit of Anaplastic Carcinoma](#)



Royal Hospital for Children, Glasgow

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[Records](#)
[Statistics](#)
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Upload Record to Database - Core Information & Diagnosis

Register ID: 2051

- Fields marked with * are mandatory to fill in.
- Only Core Information & Diagnosis, is mandatory. Other sections are all optional.

Save Core Info & Diagnosis

Core Information

Country & Centre	[?] *	United Kingdom Glasgow
Local ID	[?]	Please keep a local copy of the Registry ID you will be provided with after saving this entry. This ID will map your local patient record with the registry record.
Year of Birth	[?] *	2010
Age of First Presentation	[?]	< 1 month
Clinician	[?] *	Ahmed
Contact	[?] *	faisal.ahmed@glasgow.

DSD Classification

Sex Assigned	[?] *	☒ M ☐ F ☐ Not Assigned
Karyotype	[?] *	46 XY
Disorder Type & Actual Diagnosis	[?] *	Congenital Adrenal Hyperplasia 21-hydroxylase def (CYP21A)
Free Text	[?]	test

Consent has been obtained to share data with (*): Registered International Collaborators - Sharing data across all international partners

Save Core Info & Diagnosis

A New CAH visit

- Please note that a new CAH visit will be a copy from the last saved visit. If this is the first visit, Not Known will be default selection for Radio buttons below.
- Any change to CAH static data will also be saved when creating or updating CAH visits.

[Save CAH Visit](#)
[Next Section](#)

CAH		Blood Tests	
Date	[?] *	Test	Result
Age	[?] years	Sodium	[?] Select
		Potassium	[?] Select
		11-deoxycortisol	[?] Select
		Renin	[?] Select
Anthropometry		Adverse Events Since Last Visit	
Weight	[?] kg	Number of discrete sick day episodes [View Selected]	[?] *
Height	[?] cm	Number of sick days in total	[?]
Waist Circumference	[?] cm	Current Medication	
Hip Circumference	[?] cm	Has treatment changed	[?] * --- Please Select ---
BMI	[?]	Why was treatment changed	[?] * --- Please Select ---
BSA	[?]	Glucocorticoids [View Selected]	[?] ☐ Yes ☐ No ☐ Not Known
Cushingoid	[?] ☐ Yes ☐ No ☐ Not Known	Fludrocortisone	[?] ☐ Yes ☐ No ☐ Not Known
Virilisation	[?] ☐ Yes ☐ No ☐ Not Known	Fludrocortisone dose	[?] mcg
		Fludrocortisone frequency	[?] ☐ OD ☐ BD ☐ TDS
Blood Pressure		Salt replacement	[?] ☐ Yes ☐ No ☐ Not Known
Systolic	[?] mm Hg	Salt replacement dose	[?]
Diastolic	[?] mm Hg	Oestrogen	[?] ☐ Yes ☐ No ☐ Not Known
		Testosterone	[?] ☐ Yes ☐ No ☐ Not Known
		GnRH analogues	[?] ☐ Yes ☐ No ☐ Not Known
		Antihypertensives	[?] ☐ Yes ☐ No ☐ Not Known
		Antidiabetic	[?] ☐ Yes ☐ No ☐ Not Known
		Antidepressants	[?] ☐ Yes ☐ No ☐ Not Known
		Other drugs	[?] ☐ Yes ☐ No ☐ Not Known
		Comorbid Conditions and Surgery	
		EUA	[?] ☐ Yes ☐ No ☐ Not Known
		Surgery	[?] ☐ Yes ☐ No ☐ Not Known
		Other surgery	[?] ☐ Yes ☐ No ☐ Not Known
		Other congenital abnormalities	[?] ☐ Yes ☐ No ☐ Not Known
		Diabetes Mellitus Type 1	[?] ☐ Yes ☐ No ☐ Not Known
		Diabetes Mellitus Type 2	[?] ☐ Yes ☐ No ☐ Not Known
		Haematocrit	[?] Select
		SHBG	[?] Select
		Bone Age	
		Date of test	[?]
		Result	[?]
		Method	[?] --- Please Select ---
		Bone Mineral Density done	[?] ☐ Yes ☐ No ☐ Not Known
		Bone Mineral Density Test Date	[?]
		Bone Mineral Density Test Result	[?] --- Please Select ---
		Bone Mineral Density Test Result Note	[?]
		Male	
		Genital Stage	[?] --- Please Select ---
		Pubic hair stage	[?] --- Please Select ---
		Axillary hair stage	[?] --- Please Select ---
		Testicular Volume Left	[?] ml
		Testicular Volume Right	[?] ml
		Testicular texture	[?] --- Please Select ---
		Evidence of TART	[?] ☐ Yes ☐ No ☐ Not Known
		Fertility desired	[?] ☐ Yes ☐ No ☐ Not Known
		Attempted	[?] ☐ Yes ☐ No ☐ Not Known
		Sperm Storage	[?] ☐ Yes ☐ No ☐ Not Known
		Number of offspring	[?]
		History of assisted conception	[?] ☐ Yes ☐ No ☐ Not Known

Steering Committee

Anna Nordenstrom (Chair)	ESPE DSD WG Chair, Stockholm
John Achermann	London
Olaf Hiort	Luebeck
Rodolfo Rey	Buenos Aires
Stephanie Bernstein	Gottingen
Carol Proctor	Patient Group
Miriam Muscarella	Patient Group
Alex Springer	Vienna
Anu Bashamboo	Paris
Richard Ross	Sheffield
Amy Wisniewski	Oklahoma

<i>Project Management Group</i>		
Faisal Ahmed (PI)		University of Glasgow
Martina Rodie (Co-I)		University of Glasgow
Richard Sinnott (Co-I)		University of Melbourne
Jillian Bryce (Project Manager)		University of Glasgow
Jipu Jiang (Database Engineer)		University of Glasgow
John Watt (GRID Engineer)		University of Glasgow

- PMG meets every month
- Close links to DSDnet, ESPE DSD WG, I-CAH

- Steering Committee members serve for 3 yrs.
- Any I-DSD user can apply
- Meeting 1-2 per year



Thanks