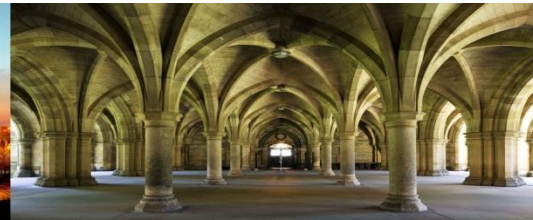


The I-DSD and I-CAH Registry Update

Jillian Bryce
I-DSD/I-CAH Project Manager

Presentation available at www.i-dsd.org



I have nothing to disclose



Evolution of the I-DSD Registry

2007-2008



ESPE
DSD
Registry

2008-2011



European
DSD Registry



2011-2016



International DSD
Registry



2014-2016

WELCOME TO I-CAH

INTRODUCTION TO I-CAH

The International-CAH network (I-CAH) is an initiative to improve the clinical management of Congenital Adrenal Hyperplasia through providing information to patients and clinicians and developing research to optimise healthcare. The I-CAH Registry is central to the I-CAH network and provides a means of connecting clinical and research centres from around the world within a Virtual Research Environment (VRE). The registry underpins research into new management strategies and therapies.

FOR PATIENTS



I-CAH provides patients with links to websites that offer a variety of support and advice to both...

[Read more »](#)

FOR PHYSICIANS



I-CAH provides clinicians with links to recent publications on the management of CAH and also web...

[Read more »](#)

REGISTRY



The I-CAH Registry is central to the I-CAH network and provides a means of connecting clinical an...

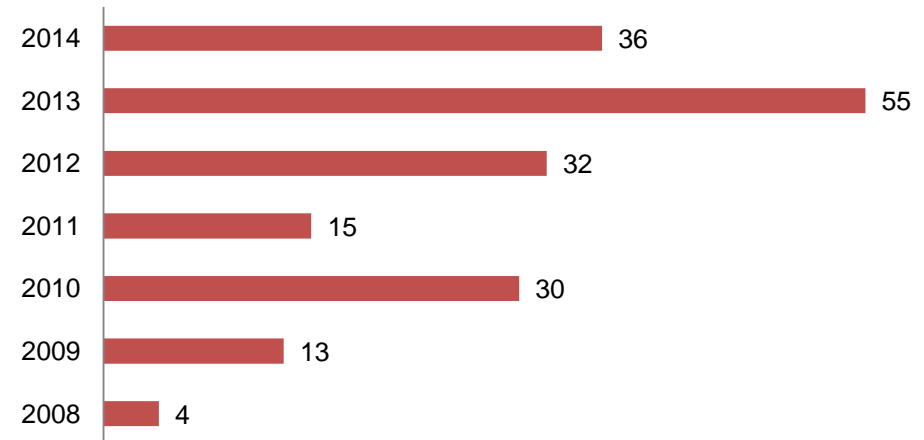
[Read more »](#)

List Of Countries By Continent

EUROPE	AFRICA	AUSTRALIA
Austria	Algeria	Australia
Belarus	Egypt	
Belgium	Malawi	
		NORTH AMERICA
Bosnia and Herzegovina	Morocco	Canada
Cyprus	Nigeria	Mexico
Czech Republic	Sudan	USA
Denmark		
Estonia	ASIA	SOUTH AMERICA
France	China	Argentina
Germany	Georgia	Brazil
Italy	Hong Kong	Colombia
Lithuania	India	
Netherlands	Indonesia	
Poland	Israel	
Romania	Jordan	
Serbia	Kuwait	
Spain	Malaysia	
Sweden	Pakistan	
Switzerland	Russia	
Turkey	Sri Lanka	
United Kingdom	United Arab Emirates	

new since Aug 2013
adding cases

Numbers of Users Registering



Registry Users Activity (185, 75% active)

- 122 Clinical Users (15 also have Researcher access)
- 4 Researchers
- 59 Basic Users

109 Centres in 47 countries

33 uploading cases in 20 countries

1387 cases (August 2014)

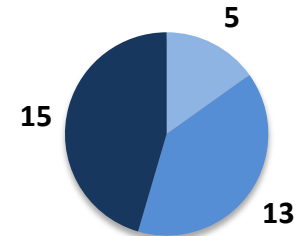
Upload rate around 100 cases per year

Main Activities

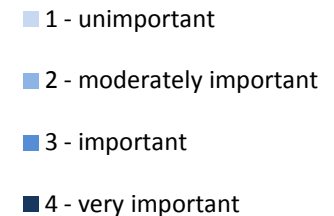
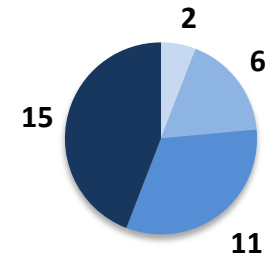
• Registry development

- Core dataset - mandatory
- CAH (live)
- Hypospadias (in final development phase)
- Patient Access to Own Record (in development)
- Transition (in development)
- Biochemical (in development)
- I-CAH Infacort study (in development)

Biochemical test



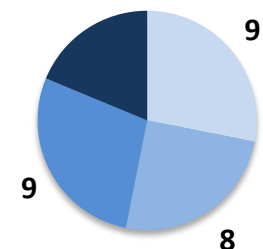
Surgical anatomical
evaluation



• Registry Governance

- New and Retiring members of the Steering Committee
- Ethics Renewal of Registry
- New SOP 3.0

Patient Access





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 TCLE GIEDDS](#)

China

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

Egypt

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

France

- [Info Eclairée Patients EuroDSD](#)

Germany

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

Israel

- [Israel DSD סופס הסכמה רישום מאגר](#)

Italy

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

Morocco

- [Morroco Copie de consentement parl](#)

Poland

- [Poland Informed Consent](#)

Romania

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

Spain

- [CI I-DSD-Unico-Final](#)
- [Spanish Information sheet for adolescents](#)

Switzerland

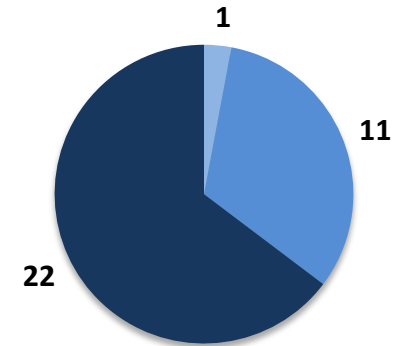
- [Switzerland 20131121_IC_DSD](#)
- [Switzerland 20131121_StudyInfo_DSD](#)

Turkey

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

- Primary research on existing cases or new cases to gather new datasets
- Secondary research on information already available
- Link to active studies on Registry Portal www.i-dsd.org to www.gla.ac.uk/idsd 'Research Studies'
- 8 active (ongoing) studies
- Outputs from recently completed studies (Incl. EuroDSD)

Database of active studies



- Cox et al 2014 JCEM 99: E348-E355

J C E M O N L I N E

A d v a n c e s i n G e n e t i c s — E n d o c r i n e R e s e a r c h

Novel Associations in Disorders of Sex Development: Findings From the I-DSD Registry

Kathryn Cox, Jillian Bryce, Jipu Jiang, Martina Rodie, Richard Sinnott, Mona Alkhawari, Wiebke Arlt, Laura Audi, Antonio Balsamo, Silvano Bertelloni, Martine Cools, Feyza Darendeliler, Stenvert Drop, Mona Ellaithi, Tulay Guran, Olaf Hiort, Paul-Martin Holterhus, Ieuan Hughes, Nils Krone, Lidka Lisa, Yves Morel, Olle Soder, Peter Wieacker, and S. Faisal Ahmed

- Kolesinksa et al 2014 *Pediatrics* 134 (3) 1-6

PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Changes Over Time in Sex Assignment for Disorders of Sex Development

Zofia Kolesinska, S. Faisal Ahmed, Marek Niedziela, Jillian Bryce, Marta Molinska-Glura, Martina Rodie, Jipu Jiang, Richard O. Sinnott, Ieuan A. Hughes, Feyza Darendeliler, Olaf Hiort, Yvonne van der Zwan, Martine Cools, Tulay Guran, Paul-Martin Holterhus, Silvano Bertelloni, Lidka Lisa, Wiebke Arlt, Nils Krone, Mona Ellaithi, Antonio Balsamo, Inas Mazen, Anna Nordenstrom, Katherine Lachlan, Mona Alkhawari, Pierre Chatelain and Naomi Weintrob

Pediatrics; originally published online August 4, 2014;

DOI: 10.1542/peds.2014-1088

- Jayasena et al 2014 Poster at ESPE: P-D-2-2-581



Scientific Programme, ePosters & Abstracts

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ESPE Abstracts (2014) P-D-2-2-581



Rare Disease Registries: Perception of Parents and Young People

Arundathi Jayasena, Miriam Muscarella & Faisal Ahmed



- Continued Development of New Modules
- Launch of Patient Access to Own Record
- Connecting with DSDnet to perform survey on clinical care
- Launch of I-CAH website
- Supporting I-CAH Infacort study
- Awarding of I-DSD Travel Grants
- Autumn Newsletter – Sept 2014
- 5th I-DSD Symposium – with COST Action DSDnet training school

5th International Symposium DSD

- 11th-13th June 2015
- Ghent, Belgium
- 18 invited speakers
- Parallel sessions
- Guest Lecture
- EACCME accreditation
- I-DSD Training Workshop
- Social Evening
- Excursion
- Discounted Registration



Thank you!

BALLOT PAPER	
Vote (X) ONLY ONCE	
	<input type="checkbox"/>
	<input type="checkbox"/>



University
of Glasgow



I-DSD Steering Committee

Ian Ford (Chair)

John Achermann

Olaf Hiort

David Sandberg

Berenice Mendonca

Claudia Wiesemann

Richard Sinnott

Miriam Muscarella

Alexander Springer

Leendert Looijenga

Amy Wisniewski

Carol Proctor

University of Glasgow

University College London

University of Lubeck

University of Michigan

University of Sao Paulo

Universität Göttingen

University of Melbourne

Harvard University

Medical University of Vienna

Erasmus MC

University of Oklahoma

Support Group Representative

Project Management Group

Faisal Ahmed (PI)

Martina Rodie (Co-I)

Jillian Bryce (Project Manager)

(University of Glasgow)

Jipu Jiang (Database Developer)

John Watt (IT security)

- meet annually (twice in year 1)
- participate in planning and oversight of I-DSD Registry
- advise its project management group
- provide suggestions on its future plans
- ensure I-DSD objectives are being adequately addressed
- 2-3 year term – will need different disciplines
- **oversee the approval of proposed research studies**

- **An effective understanding of the aetiology of any rare condition as well as long term outcome requires multicentre collaboration across national boundaries.**
- **The I-DSD Registry consists of brief non-identifiable details of over 1000 affected people with a range of rare conditions leading to a DSD.**
- **Clinicians involved in the care of a person with a rare condition can find details of other specialists with experience of the same condition**
- **Registered users can use information in the registry to design new studies and recruit to existing studies**
- **The I-DSD Registry acts as a secure clinical and research network and adheres to the highest standards of data governance and security**

User account management

Clinical User

- Can upload cases.
- Can view whole dataset of own cases and core dataset of all other cases.
- Only centre lead can delete cases from their centre.

Research User

- Can view whole dataset of all cases once approved.
- Research access applied for a fixed 6 month period.
- Access renewed upon submission of a report

Basic User

- Can't view any case data.
- Can search other user's profiles.

Accounts that are inactive for 12 months or more will be removed.

Research Studies - list

Patient Information Sheets in multiple languages

Surveys

Publications

DSD net

5th I-DSD Symposium

Upload rates

Newsletter

I-CAH longitudinal study

Patient Access

Steering Committee

I-DSD Travel Grants

First presentation: <1 month - 53 y, median 1-3 months, median yob 1998 (range 1927-2014)

Majority of cases: 46XY karyotype (n=983), then 46XX (n=258)

Sex assigned: female 55% (n=764), male 45% (n=621)

25 males with 46XX karyotype, 483 females 46XY karyotype

	<u>% of cases</u>
Associated conditions	25% (n=348)
History of infertility	7% (n=93)
History of DSD	19% (n=263)
Parental consanguinity	13% (n=178)
Mosaicism	5% (n=71)
Samples available	39% (n=546)

I-DSD Travel Grants

- maximum of £500
- cover travel, accommodation and conference fees
- can be used to:
 - present an oral or poster at a recognised conference or similar on research using the I-DSD registry
 - Finance a short-term visit to a suitable host institute/laboratory to work on research using the I-DSD registry
 - Collaborate with colleagues in another location to develop modules for use in the I-DSD registry
 - Test samples available through the I-DSD registry in the framework of a joint research project.
- awarded ad hoc (apply anytime)
 - forms available from www.gla.ac.uk/idsd



1387 cases (August 2014)

Upload rate around 100 cases per year

Commonest disorder type: Disorders of androgen action (n=382)
followed by: Disorders of gonadal development (n=304)

First presentation: <1 month - 53 y, median 1-3 months, median yob 1998 (range 1927-2014)

Majority of cases: 46XY karyotype 71% (n=983), then 46XX 19% (n=258)

Sex assigned: female 55% (n=764), male 45% (n=621)

25 males with 46XX karyotype, 483 females with 46XY karyotype