**I-DSD Steering Committee Update 8th June 2015**

Regarding the I-DSD registry, there are several matters arising that we want to inform the Steering Committee of and seek feedback/advice.

**A. Research Studies**

Update on three studies which have recently completed:-

1. Kathryn Cox (Glasgow) (Feb 12-Jun 14): Incidence of congenital anomalies associated with DSD (completed). Output: Oral and poster presentations at BES (Harrogate, March 2013), I-DSD symposium (Glasgow, June 2013), ENDO (San Francisco, June 2013), ESPE (Milan, Sept 2013) Publications: Endocrine Abstracts (2013) 31 P148 DOI:10.1530/endoabs.31.P14. Cox et al. Novel associations in disorders of sex development: findings from the I-DSD Registry.   J Clin Endocrinol Metab, February 2014, 99(2):E348–E355 [JCEM jc.2013-2918; doi:10.1210/jc.2013-2918](http://jcem.endojournals.org/content/early/2013/12/03/jc.2013-2918.full.pdf%2Bhtml)
2. Zosia Kolesinska (Poznan) (Sep 13-Mar 14): looking at trends in sex assignment.

Output: Oral presentation at I-DSD symposium (Glasgow, June 2013), ESPE (Milan, Sept 2013). Publication Kolesinska et al. Temporal Changes In Sex Assignment In Disorders Of Sex Development – Lessons From The International DSD Registry. Pediatrics (2014) doi: 10.1542/peds.2014-1088

1. Arundathi Jayasena (Colombo) & Miriam Muscarella (Boston) (Jun-Nov 13): Perception of rare disease registries by parents and young people. Output: Poster ESPE (Dublin, Sept 2014) <http://espe2014abstracts.eurospe.org/hrp/0082/eposters/hrp0082p2-d2-581_eposter.pdf>
2. Angela Lucas-Herald (Glasgow) (Dec 13-Feb 15): Outcome in male PAIS. Long Oral at I-DSD symposium 2015 (Ghent)

Update on studies which are underway:

1. John Achermann (London) (Mar 13- open): Novel mechanisms in adrenal and reproductive biology. Initial focus on own centre, will begin recruitment from I-DSD Registry in early 2014.
2. Martine Cools (Ghent) (Jan 13-open) Outcome of preserved gonads in adults with AIS. Questionnaires developed and study underway. Complete in 2015.
3. Guilherme Guaragna Filho (Campinas) (Mar-14-open): Outcome in female PAIS. Presentation at I-DSD symposium 2015.
4. Nanjananya Karunasena & Richard Ross (Sheffield) (May 15- open): Prevalence and epidemiology of CAH in I-DSD registry
5. Andreas Kyriakou (Glasgow) (Sept 14-open) COST DSDnet WG4 survey Output: 2 Oral and 1 poster presentations at I-DSD symposium 2015.
6. Sukran Poyrazoglu & Feyza Darendeliler (Istanbul) (Feb 15-open) Birth weight in Different Etiologies of DSD
7. Ken McElreavy (Paris) (May 2014-open) Exome sequencing of DSD
8. Richard Ross (Sheffield) (Jan 14-open) CAH in adults – launched I-CAH registry – planning clinical trials

The following have been assisted with developing their studies and grant applications

1. Filiz Mine Cizmecioglu (Koecali) (Feb 15-open) CAH prevalence in Turkey (in development)
2. Nils Krone (Birmingham) CAH in children – using I-CAH registry to look at long-term outcomes in CAH (in development)
3. Marie Lindhart Johansen & Anders Juul (Copenhagen) - Outcome in X/XY boys and men (in development)
4. Mohamed A Baky Fahmy (Cairo) Androgen Receptor Deficiency in Rare Genitourinary Anomalies (in development)

Other possible studies

1. Leendert Looijenga (Rotterdam) & Martine Cools (Ghent) - Ovotesticular cases of DSD on the registry (in discussion)
2. Nils Krone (Birmingham) CAH-UK – grant under review (discuss in Ghent)

*Dormant Studies*

1. *Birgit Kohler (Berlin) (Aug 13-Jun 14): DSD life – a Clinical European study investigating the care and therapies of DSD patients. (Not used Registry - last login Nov 2012)*
2. *Ediz Yeşilkaya (Ankara) (Dec 13-Jun 14): Chronic complications in girls and women with Turner syndrome. (No updates – last login Nov 13)*
3. *O Evliyaoglu (Istanbul) (Mar 13-Sept 13): DSD, clinical and molecular pathogenesis – Research User from March 2013. (No updates – last login Jan 14)*
4. *Vicky Pasterski (Cambridge) Gender identity in AIS – (Not funded)*

**B. I-DSD Registry modules**

The I-DSD Registry database is a modular based interface.

The core dataset is mandatory for each record added and there are currently 4 additional optional modules, with scope to develop further specific modules (surgical anatomical, biochemistry, transition).

On 20th May 2015, the longitudinal module for CAH was launched. This was developed to support the EU-TAIN project for CAH medication (R Ross, Sheffield).

Future directions include providing modular access to researchers, developing a fee structure for obtaining access, personalising the page viewed by patient participants.

**C. I-CAH Website**

A new I-CAH website was launched in October 2015 ([www.i-cah.org](http://www.i-cah.org)). The website provides information on CAH care to physicians and patients and provides a portal to the I-CAH registry which is hosted by the I-DSD registry. ([www.registry.i-cah.org](http://www.registry.i-cah.org))

**D. User roles and access**

In the past year we changed the level of access users have in order to monitor Registry function as a platform for clinical networking and research. All active registered users are able to access some of the core data. In addition, those who are contributing cases can access all the information on their cases. Access to more information on other cases is provided for limited periods and for specific research projects. Users who have become inactive (i.e. not logged in for 12 months or more) have had their access to data suspended. These user’s profiles are retained in the User database for networking purposes. Information on the various roles and how to register for research access are available on the I-DSD website ([www.gla.ac.uk/idsd](http://www.gla.ac.uk/idsd)). Research users need to provide a 6-monthly report or their access is suspended.

**E. Patient Access**

Registry participants or their guardian (if under 16) are now able to access a portion of their record on the I-DSD Registry.  Access to the anonymised data is via a secure login page on the Registry website ([ww.i-dsd.org](http://www.i-dsd.org/)).  Access is set up according to step-by-step guidelines available on the website.

**F. Terms and Conditions**

These are covered in version 4 of the Standard Operating Protocol (on [I-DSD website](http://www.gla.ac.uk/schools/medicine/research/childhealth/i-dsdproject/thei-dsdregistry/standardoperatingprotocol/))

**G. I-DSD Symposium**

The 5th I-DSD symposium will be held in Ghent on 11th-13th June 2015. It is combined with the DSDnet training school. Martine Cools, Piet Hobeke and Leendert Looijenga are the local organisers with support from the I-DSD project management office. Total number of delegates registered by 4th June 2016 were 219.Almost 80 abstracts were submitted and in addition to the invited speakers there will be 25 oral presentations and 50 posters. The I-DSD Training Workshop is fully subscribed with 54 participants undergoing e-learning, communications and I-DSD registry training.

**H. I-DSD Support Grants**

The 4th I-DSD symposium generated a small surplus of funding which the Steering Committee agreed could be used to promote the use of the I-DSD Registry. These funds have been used to support travel for individuals who use the I-DSD registry for research, including giving a presentation of an I-DSD research study or to visit a centre to work on an I-DSD Registry related project (modules, patient access, website development, etc). Grants are limited to £500 (equivalent).

We welcome volunteers from the Committee to become involved in the review process of this grant scheme or make suggestions on other ways to use these funds.

**I. Steering Committee Tenure**

Members who have demitted their post in the committee to date are: Ieaun Hughes, Feyza Darendililer, Ellie Magritte and David Sandberg. In the past year the committee was joined by new members; Amy Wisniewski, Richard Ross and Carol Proctor. Members to demit in the forthcoming year are: Berenice Mendonca, Claudia Wiesemann and John Achermann (Jun), Alex Springer, Miriam Muscarella and Leendert Looijenga (Oct), Olaf Hiort (Nov). The Steering Committee is invited to suggest names of possible future committee members. Ken McElreavy (Paris) and Selma Witchel (Pittsburgh) are already on the list.

**J. Steering Committee Meetings**

We intend to hold the next annual meeting in June 2016 possibly by teleconference or in person at ESPE in Paris, Sept 2016. Thoughts please?

**K. DSDnet**

COST action BM1303 DSDnet, chaired by Olaf Hiort, was launched in mid 2014. There have been several working group meetings, a round of STSMs (short term scientific missions) and most recently the 1st Training School in Ghent June 2015. Further details of the action can be accessed at [www.dsdnet.eu](http://www.dsdnet.eu). The I-DSD Registry is closely linked to this action – through WG4 - and the two complement each other.

**L. NICHD Workshop ‘Growing Up with DSD’**

NICHD in the USA held a Workshop on DSD supported by the Endocrine Society in Bethesda in March 2014.  The purpose and scope included identifying gaps in knowledge about the effects of DSD in affected individuals and above all, to identify research questions that will lead to a scientific evidence base.  The Workshop was not intended as another Consensus meeting or to issue Guidelines. A multi-disciplinary group of clinical and research experts and members of advocacy groups will participate.  The participants included two members of the I-DSD Steering Committee.- David Sandberg and Ieuan Hughes. The outcome was a special issue of *Hormone and Metabolic Research* comprising articles reflecting proceedings at the workshop which was published in May 2015. HMR have very generously provided 125 copies of the special issue for distribution at the I-DSD symposium in Ghent.

**M. Rare Disease Registries**

The EPIRARE project ([www.epirare.eu](http://www.epirare.eu)) that was looking at an EU Platform for Rare Diseases is now completed. Through its activities with its members over the past 3 years, its final deliverables were published on its website in March 2014.

Epirare co-ordinates the workpackage on RD registries in RD-Connect, (an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research) and plans to launch a catalogue of RD-Connect registries and biobanks, which will be a gateway towards data sharing of different data sources.

**N. Policy on surveys**

I-DSD wants to increase the use of the Registry as a tool for clinical interaction. If an I-DSD user receives a personal email from another I-DSD user asking them about their practice or experience of a certain condition, that should be considered a clinical query and not a survey.  Such a query has not been officially approved by I-DSD and it is up to the user to respond as they see fit.

Surveys of the I-DSD user community are welcome but are subject to the same application process as for [Research Studies](http://www.gla.ac.uk/schools/medicine/research/childhealth/i-dsdproject/research/).  Taking part in a survey is entirely optional for I DSD Users. However, if the survey requires approaching the actual Registry participant whose data have been entered into the Registry, the survey will be considered a study that requires separate ethics approval.

Emails that are seeking participation in an official survey approved by I-DSD will only be issued from the I-DSD office.

**O. Other planned activities**

A spring newsletter was issued in March 2015 highlighting some of the above activities. The next newsletter will be in Autumn prior to ESPE. Please send any items by mid-August

If anybody wants more details on any of these projects or wants to be involved, then please get in touch with me.

Best Regards

Jillian Bryce