**I-DSD Steering Committee Update 12th March 2014**

Regarding the I-DSD project, 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) – Incidence of congenital anomalies associated with DSD (completed).

Output: Oral and poster presentations at BES (Harrowgate, March 2013), I-DSD symposium (Glasgow, June 2013), ENDO (San Francisco, June 2013), ESPE (Milan, Sept 2013). Publication 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+html)

1. Zosia Kolesinska (Poznan) - looking at trends in sex assignment (completed).

Output: Oral presentation at I-DSD symposium (Glasgow, June 2013), ESPE (Milan, Sept 2013). Manuscript Kolesinska et al. Temporal Changes In Sex Assignment In Disorders Of Sex Development – Lessons From The International DSD Registry. Submitted for peer review February 2014.

1. Arundathi Jayasena (Colombo) & Miriam Muscarella (Boston) Perception of rare disease registries by parents and young people – Submitted as an abstract to ESPE

Update on studies which are underway:

1. John Achermann (London) Novel mechanisms in adrenal and reproductive biology. Research User from March 2013. Initial focus on own centre, will begin recruitment from I-DSD Registry in early 2014.
2. Birgit Kohler (Berlin) intends to use the Registry for DSD life – a Clinical European study investigating the care and therapies of DSD patients. Research user from August 2013. Not used Registry
3. O Evliyaoglu (Istanbul). DSD, clinical and molecular pathogenesis – Research User from March 2013. Awaiting 6-month report.
4. Angela Lucas-Herald (Glasgow). Outcome in male PAIS –. Research User from Dec 2013 - data collection is ongoing
5. Ediz Yeşilkaya (Ankara). Chronic complications in girls and women with Turner syndrome –Research User from Dec 2013. Awaiting update.
6. Guilherme Guaragna Filho (Campinas) Outcome in female PAIS. – Research User from March 2014

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

1. Martine Cools (Ghent) - Outcome of preserved gonads in adults with AIS – (in development)
2. Vicky Pasterski (Cambridge) - Gender identity in AIS – (in development)
3. Richard Ross (Sheffield) CAH in adults – launched I-CAH registry – planning clinical trials (in development)
4. Nils Krone (Birmingham) CAH in children – using I-CAH registry to look at long-term outcomes in CAH (in development)

Other possible studies

1. Anders Juul (Copenhagen) - Outcome in X/XY boys and men (in discussion)
2. Leendert Looijenga (Rotterdam) & Martine Cools (Ghent) - Ovotesticular cases of DSD on the registry (in discussion)

**B. I-DSD Registry modules**

The I-DSD Registry database has undergone a major revision in the past year and is now a modular based interface. The core dataset is mandatory for each record added and there are currently 4 additional optional modules (including the recently launched CAH module) with scope to develop more as needs arise. E.g.

* Surgical Anatomical module (Alex Springer, Vienna) – Hypospadias.
* A module that captures biochemical data (previously discussed with SC)
* Transition module (Helena Gleeson, Leicester) to develop a method of capturing activity that is related to transfer of patients to adult care.
* Longitudinal module for CAH medication (Richard Ross, Sheffield)

**C. I-CAH Registry**

The I-CAH registry was launched on March 3rd 2014. This registry is hosted by the I-DSD registry and is essentially the same database behind a different front portal ([www.-i-cah.org](http://www.-i-cah.org)). The I-CAH Registry SOP and Patient consent forms have been given Ethics Approval and recruitment is now underway. Jillian will act as manager of the I-CAH Registry part-time supported by separate funding from I-CAH.

**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. Terms and Conditions**

We are currently seeking legal advice on generating Terms and Conditions for using the Registry which includes clarification on aspects of ownership of data. Our recommendation is that the data are owned by the following in order of priority - the patients themselves; the lead for the centre that are uploading data; the registry host (University of Glasgow). The new T&Cs will also clarify the guidelines for authorship of papers that involve research using the registry data with the recommendation to follow the guidelines of ICMJE (<http://www.icmje.org/>).

**F. I-DSD Symposium**

The 5th I-DSD symposium will be held in Ghent on 11th-13th June 2015. It is likely that this will be combined with a DSDnet training school. Martine Cools, Piet Hobeke and Leendert Looijenga are the local organisers with support from the I-DSD project management office. The structure of the symposium is being shaped and invited speakers identified. As with the previous symposium, a workshop for new investigators is proposed as well as parallel session for patients, paramedical and surgeons. A provisional programme is attached to the SC for comment before speakers are invited

**G. I-DSD Support Grants**

In a previous e-mail, the Steering Committee were informed of a small surplus of funding available through income from the Glasgow I-DSD symposium. To promote the use of the I-DSD Registry, these funds will be used to support travel for individuals who use the I-DSD registry for research. Possibilities include 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 will be limited to £500 (equivalent).

We welcome volunteers from the Committee to become involved in the review process of this grant scheme.

**H. Steering Committee Tenure**

Members who will demit their post in the committee in 2014 are:  Ieaun Hughes, Feyza Darendililer and Ellie Magritte (all May) and David Sandberg (November). The committee was recently joined by new members, Leendert Looijenga, Alex Springer and Miriam Muscarella. Others who have been identified for the future include: Amy Wisniewski (Oklahoma City) (May) and Richard Ross (Sheffield) (Nov), Ken McElreavy (Paris), Carol Proctor (Parent) and Selma Witchel (Pittsburgh).

**I. Steering Committee Meetings**

We intend to hold the next annual meeting in June 2014 (1 year on from the last meeting in Glasgow). It is proposed that this will be a telephone conference and a doodle poll will be circulated. There are also opportunities for some members of the committee to meet informally at upcoming conferences, such as ENDO in Chicago in June and ESPE in Dublin in September.

**J. DSDnet**

Olaf Hiort is chairing a new COST action on DSD. Further details of the action can be accessed at [www.cost.eu/domains\_actions/bmbs/Actions/BM1303](http://www.cost.eu/domains_actions/bmbs/Actions/BM1303). The I-DSD Registry will be closely linked to this action and the two will complement each other.

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

NICHD in the USA is holding a Workshop on DSD supported by the Endocrine Society.  The purpose and scope includes 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 is 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 include two members of the I-DSD Steering Committee.- David Sandberg and Ieuan Hughes.

**L. 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 close to completion. Through its activities with its members over the past 3 years, its final deliverables will published on its website in March 2014

**M. Other planned activities**

A spring newsletter will be issued in March 2014 and will highlight some of the above activities.

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