

I-DSD/I-CAH newsletter - August 2018

Welcome to the August newsletter

This is the 4 monthly update on current activities in the I-DSD and I-CAH registries. If you have an item you would like to include in future newsletters, please contact Jillian.Bryce@glasgow.ac.uk



I-DSD/I-CAH user group

Athens

26 September 2018

FREE



The next I-DSD/CAH user group meeting will take place at Sofia Children's Hospital in Athens on 26th September 2018 between 5-9pm.

Attendance is free but please [register on-line](#) by 31st August. There is an option to join us for a meal afterwards.

Further information including the final programme is available from the [event website](#)

User membership

As the I-DSD/I-CAH Registries become independent of any single anchor funder, they are adopting a new membership structure with different levels of membership. For full details regarding membership costs and benefits, please see [here](#).

I-DSD 2019

Sao Paulo

4-6 July 2019



The next I-DSD Symposium will take place in Sao Paulo, Brazil from 4-6th July 2019.

The programme is now available on the [I-DSD 2019 website](#). The programme follows the format of previous symposia. Registration and abstract submission will be available in 2019.

I-DSD/CAH Registries Related Outputs Over The Last Year

For full author details, please see the [list of publications](#) on the website.

Sanders C et al. 'Involving individuals with disorders of sex development and their parents in exploring new models of shared learning: Proceedings from a DSDnet COST action workshop'. *Sex Dev* 2018 Jun 23. doi: 10.1159/000490081.

[Recommendations for Improving the Quality of Rare Disease Registries](#). Kodra Y, Weinbach J, Posada-de-la-Paz M, Coi A, Lemonnier SL, van Enckevort D, Roos M, Jacobsen A, Cornet R, Ahmed SF, Bros-Facer V, Popa V, Van Meel M, Renault D, von Gizycki R, Santoro M, Landais P, Torreri P, Carta C, Mascalzoni D, Gainotti S, Lopez E, Ambrosini A, Müller H, Reis R, Bianchi F, Rubinstein YR, Lochmüller H, Taruscio D. *Int J Environ Res Public Health*. 2018;15(8).

Kourime M et al. 'Virtual Networks for Exchanging Information and Biomaterials: Future Directions'. *Sex Dev*. 2018;12: 134-138.

Poyrazoglu S et al. 'Birth weight in different etiologies of disorders of sex development'. *J Clin Endocrinol Metab*. 2017; 102:1044-1050.

Kourime M et al. 'An assessment of the quality of the I-DSD and the I-CAH registries - international registries for rare conditions affecting sex development'. *Orphanet J Rare Dis*. 2017; 12:56.

Dessens A et al. 'Understanding the needs of professionals who provide psychosocial care for children and adults with disorders of sex development'. *BMJ Paediatrics Open*, August 2017 .

Kodra Y et al. 'Data Quality in Rare Diseases Registries'. *Adv Exp Med Biol*. 2017; 1031:149-164.

Active Research Studies

Jeremy Tomlinson (Oxford), 'Optimizing mineralocorticoid replacement in patients with CAH'

Katya DeGroot (Ghent), 'Cardiovascular pathology in patients with 45,X/46,XY (or variant) karyotypes'

Lloyd Tack (Ghent), 'Growth and pubertal course in 46,XY SGA boys born with atypical genitalia of unknown origin'

Angela Lucas-Herald (Glasgow), 'Trends in gonadectomy in DSD'

Ken McElreavey (Paris), 'Defining new genetic aetiologies for syndromic DSD'

Alexander Springer (Vienna), 'Changes in surgical practice in CAH'

Nadine Hornig (Kiel), 'Identification of co-regulators of the androgen receptor leading to AIS type'

Nils Krone (Sheffield), 'Defining the dose, type and timing of glucocorticoid treatment in children and adults with Congenital Adrenal Hyperplasia (CAH-UK)'

Recently Completed Studies:

Hedi Claahsen & Uta Neumann (Nijmegen & Berlin), 'Fludrocortisone study in <3year olds with CAH'. In final analysis'

Lloyd Tack, Martine Cools (Ghent), 'Management of gonads in adults with androgen insensitivity: an international survey'

Marie-Lindhart Johansen, Anders Juul (Copenhagen), 'Long-term outcomes in males with 45X/46XY mosaicism'

Guilherme Guaragna Filho (Campinas), 'Evaluation of Puberty in Females with Partial Androgen Insensitivity Syndrome (PAIS)'

Data Sharing Agreement

Accessing data in the I-DSD and I-CAH registry is subject to completion of a [data sharing agreement](#) by the requesting party and the Office for Rare Conditions.

New Scientific Panel

We are pleased to announce that there is a new panel of experts that has been created to assess requests for accessing data from the Registry. This will consist of Faisal Ahmed (Glasgow), Anna Nordenstrom (Stockholm), Jillian Bryce (Glasgow), Salma Ali (Glasgow), Hedi Claahsen (Nijmegen), Nils Krone (Sheffield), Cunyi Wang (Glasgow), Nadine Hornig (Kiel).

Patient Area - Local Language

Using a translation tool, patients who have been given access to their record in the registry will be able to view this and their centre's details in another language. For more information on patient access – [link to website protocol](#).

Patient Advisor On Steering Committee

We have a vacancy for a patient advisor on the steering committee. Please contact Jillian.Bryce@glasgow.ac.uk