

I-DSD & I-CAH Registries Newsletter

Autumn 2016



How can the Registry help improve care?

- The Registry consists of non-identifiable details of affected people in a secure clinical and research environment
- The Registry contains contact details of clinical experts and active researchers in the field of DSD/CAH
- Users can use this information to perform studies and discuss rare conditions with other experts
- Patients can access a space that provides details of their condition and their centre

International DSD Symposium 2017



Registration opens on 1st October 2016 for the 6th I-DSD symposium which will take place from 29th June–1st July 2017 in Copenhagen, Denmark. [I-DSD symposium 2017](#)

Changes To Access Of Data In Registries

Searching the Registries for data at centres other than the user's own centre is now performed by the project management team. Please contact Jillian Bryce for further information if you would like to perform any searches.

Research Support Grants

Small grants are available now to support travel related to Registry-based research. Visit the [I-DSD website](#) or contact [Jillian Bryce](#) for more information.

Steering Committee & Project Management Group

The [I-DSD & I-CAH Registries Steering Committee](#) thanks Carol Proctor for their valuable contribution. Applications for new members of the Steering Committee are invited from current registry users.

Personalised Patient & Parent Participant View

Access for patient and parent participants can be personalised with information about the clinical centre from where the participant is registered. Centre leads are encouraged to maintain up to date information on their centre for the participant. To provide access to patients and parents see step-by-step instructions at [Participant Access](#)

ERN for rare endocrine conditions incl DSD & CAH

The application for European Reference Network for rare conditions has been submitted. I-DSD or I-CAH users who would like more information should contact [Olaf Hiort](#)

Response to the Council of Europe Human Rights

Commissioner's Issue Paper on Human Rights and Intersex People. M Cools, M Simmonds, S Elford, J Gorter, S. F.Ahmed, F. D'Alberton, A. Springer, O. Hiort *Eur Urol.* 2016 May 19. pii: S0302-2838(16)30179-8.

News From The I-CAH Registry

Well done to the CAH User Group for continuing to enter CAH cases. Over 30% of the cases in the two registries have a diagnosis of CAH. Data from the registries reveals a temporal shift in diagnostic practice of CAH towards the use of molecular genetics. Abstracts have been submitted to ESE & ESPE. Funding has been secured to develop the Registries further including a new study exploring patient reported outcomes in CAH (PRO-CAH). Further details of active studies are available at [Research Studies](#)

News From The I-DSD Registry

Well done to I-DSD users who joined forces for the following two important publications this year in the field of AIS:-

- Lucas-Herald A, Bertelloni S, Juul A, Bryce J, Jiang J, Rodie M, Sinnott R, Boroujerdi M, Lindhardt-Johansen M, Hiort O, Holterhus PM, Cools M, Guaragna-Filho G, Guerra-Junior G, Weintrob N, Hannema S, Drop S, Guran T, Darendeliler F, Nordenstrom A, Hughes IA, Acerini C, Tadokoro-Cuccaro R, Ahmed SF. *The Long Term Outcome Of Boys With Partial Androgen Insensitivity Syndrome And A Mutation In The Androgen Receptor Gene.* *J Clin Endocrinol Metab.*

- Hornig NC, Ukat M, Schweikert HU, Hiort O, Werner R, Drop SLS, Cools M, Hughes IA, Audi L, Ahmed SF, Demiri J, Rodens P, Worch L, Wehner G, Dunstheimer D, Müller-Roßberg E, Reinehr T, Hadidi AT, Eckstein AK, van der Horst C, Seif C, Siebert R, Ammerpohl O, Holterhus P-M. *Identification Of An AR-Mutation Negative Class Of Androgen Insensitivity By Determining Endogenous AR-Activity.* *J Clin Endocrinol Metab.*

Other Research Highlights

Active research studies are listed on the [I-DSD website](#)

- 33 users from 12 countries are currently developing or performing studies using the Registry

- Further details on research outputs can be accessed at [Publications & Dissemination](#)

Registry Statistics (August 2016)

- 2364 cases, uploaded by 60 centres in 27 countries over 5 continents

- Highest number of new cases added over the last 6 months was from Istanbul University (125 uploads)!

To find out more contact

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