

I-DSD/I-CAH newsletter - December 2018

Welcome to the December newsletter - revised

REVISION 19.12.18 - Apologies - an item was missing from the previous mailing. 'A New Therapy for Adult CAH: Clinical Trial' now included.

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 registry@i-dsd.org



I-DSD 2019 Sao Paulo 4-6 July 2019



I-DSD Symposium 2019

The 7th I-DSD Symposium will take place in Sao Paulo, Brazil from 4-6th July 2019.

The programme is available on the I-DSD and the [I-CAH websites](#). The programme follows the format of previous

symposia.

Registration is now open idsd2019.eventbrite.co.uk and abstract submission will open in early 2019.

CME accreditation (15 hours) has been applied for (UEMS-EACCME)

We would like to extend our thanks to Diurnal Ltd, Sandoz and ESPE for supporting the I-DSD meeting.

I-DSD/I-CAH user group meeting, Athens 26th September 2018

The I-DSD/CAH user group meeting took place at Sofia Children's Hospital in

Athens on 26th September 2018. The meeting was attended by around 150 people from 72 institutes in 30 countries. The meeting was aimed at clinical and basic scientists who use (or plan to use) the I-DSD/I-CAH registries to conduct or contribute to research. Talks on completed and ongoing studies were well received and sparked interesting discussions. Further information about this and future meetings is available on 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](#).

Outputs from the Registry over the last year

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

- Tack L et al. Management of gonads in adults with androgen insensitivity: an international survey. *Horm Res Paediatrics* 2018 doi.org/10.1159/000493645
- Ali S et al. The current landscape of European registries for rare endocrine conditions. *Eur J Endocrinol*. 2018 Nov 1. doi.org/10.1530/EJE-18-0861.
- 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.org/10.1159/000490081.
- Kodra Y, et al. Recommendations for improving the quality of rare disease registries. *Int J Environ Res Public Health*. 2018;15. doi.org/10.3390/ijerph15081644.
- Kourime M et al. Virtual networks for exchanging information and biomaterials: future directions. *Sex Dev*. 2018;12: 134-138. doi.org/10.1159/000486872
- Kodra Y et al. Data quality in rare diseases registries. *Adv Exp Med Biol*. 2017;1031:149-164. DOI.org/10.1007/978-3-319-67144-4_8.

Active Studies

- Irina Bacila (Sheffield) - Rare forms of Congenital Adrenal Hyperplasia
- Grit Sommer (Bern) - SF1 next
- Salma Ali (Glasgow) - Adverse Events in Congenital Adrenal

Hyperplasia

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

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.

The Use Of Data From The Registry

The data in the Registry are becoming increasingly attractive to a wide range of stakeholders. Clinicians looking after people with DSD find it a useful way of securely archiving the cases in a standardized way that is internationally acceptable. Academic researchers are using the data for a range of studies and more recently we have also started to interact with industry who are interested in the real world data that the Registry collects as this helps them in developing new drugs as well as understand pathways for pharmacovigilance.

All these activities are important and endorsed by regulators such as the EMA as well as patient groups. The future direction of the Registry is to develop clinical benchmarks and tools that will also help with service improvement. All data from the Registry are only shared with researchers after a thorough assessment by the Scientific Panel.

A New Therapy for Adult CAH: Clinical Trial

Spruce Biosciences is planning to conduct clinical trials in adult classic CAH patients to evaluate the safety and efficacy of SPR001 an investigational new therapy for CAH. SPR001 is a novel CRF1 receptor blocker that reduces overproduction of ACTH and adrenal hormones, leading to potential improvements in symptoms and the ability to reduce glucocorticoid use.

We plan to initiate these global clinical trials in 2019.

If you have adult CAH patients and an interest in clinical trials, please reach out to David Moriarty at dmoriarty@sprucebiosciences.com or call +1 650 862 9761. You can learn more about Spruce at <https://www.sprucebiosciences.com/>

Scientific Panel

Since its introduction in early 2018, requests for access to data from the Registry have been considered and approved by the Scientific Panel.

The Panel has also guided the researchers in refining their studies. The panel comprises Hedi Claahsen (Chair, Nijmegen), Anna Nordenstrom (Stockholm), Nils Krone (Sheffield), Nadine Hornig (Kiel) and members of the project management group in Glasgow including Faisal Ahmed (Glasgow), Jillian Bryce (Glasgow), Salma Ali (Research Fellow, Glasgow) and Adalia Ikiroma (Statistician, Glasgow).

The Steering Committee

The Steering Committee has approached European Society of Paediatric Endocrinology (ESPE), Pediatric Endocrine Society (PES), Australasian Paediatric Endocrine Group (APEG) and Asia Pacific Paediatric Endocrine Society (APPES), regional societies who have a DSD working Group for representation on the Steering Committee. Please visit the website if you are interested in joining the Steering Committee

Patient Advisor On Steering Committee

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

IFCAH news

The IFCAH scientific committee is dedicated to select the best projects worldwide that will benefit the patients and families affected by congenital adrenal hyperplasia. Since 2018 a total of 26 out of 112 projects were funded from an allocation of €2.2M.



For more information read the [2018 newsletter](#)



Season's Greetings

On behalf of the whole I-DSD/I-CAH team, thank you for your continuous support and contributions.

Season's greetings from everyone here at I-DSD/I-CAH and wishing you all a prosperous new year!

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