

# **GrApSIA y el Proyecto COST**

**Laura Audí**

**Marta Rozas**

# Proyectos Europeos colaborativos entre profesionales del ámbito científico-sanitario y asociaciones de afectad@os

Están en curso actualmente en Europa dos Proyectos dedicados a DSD.

- El primero es **DSD-Life del Programa FP7 (16) (2012-2016)**.

Reúne a 15 Grupos de investigadores europeos que incluyen endocrinólogos, psicólogos, cirujanos, ginecólogos y eticistas. El objetivo de este Proyecto es **mejorar el manejo clínico de los pacientes con DSD**. Analizan áreas de gran importancia para la calidad de vida: **calidad de vida general y bienestar psicosexual, desarrollo psicosexual, calidad y satisfacción de los tratamientos recibidos**.

El Proyecto se orienta especialmente hacia los **puntos de vista de los pacientes, la ética y los contextos culturales**. Están **integradas Asociaciones de Personas Afectadas** con el fin de incluir los puntos de vista, opiniones y propuestas de afectados y padres. Están realizando encuestas en los centros participantes, en las Asociaciones de Personas Afectadas y en el I-DSD Registry. Los resultados permitirán una **evaluación y la elaboración de nuevas Guías Clínicas**, seguidas de la diseminación de los resultados a través de las sociedades profesionales, las publicaciones científicas y los Grupos de Apoyo a las Personas Afectadas. Esta diseminación incrementará el **conocimiento del público en general sobre los DSD y su integración**.

# Proyectos Europeos colaborativos entre profesionales del ámbito científico-sanitario y asociaciones de afectad@os

- El segundo proyecto es **COST (European Cooperation in Science and Technology) BM1303, DSD-net "A systematic elucidation on Differences of Sex Development"** del Programa Horizon 2020 (2013-2017).
- Está integrado por **Grupos de 22 países europeos** (entre ellos España), **2 países vecinos** (Egipto y Rusia) y **7 Grupos internacionales** (3 australianos, 2 japoneses, 1 indonesio y un norteamericano).
- Funcionan **5 Grupos de Trabajo**:
  - Grupo 1 "**Armonización y estandarización del fenotipado clínico y del tratamiento**"
  - Grupo 2 "**Biología y Genética**"
  - Grupo 3 "**Armonización de la exploración de laboratorio**"
  - Grupo 4 "**Experiencias y Percepciones de la Investigación**"
  - Grupo 5 "**Diseminación y capacitación**"
- En algunos **Grupos de Trabajo (4 y 5)** se han incorporado personas afectadas y padres procedentes de **Grupos de Apoyo**.
- Cada Grupo está desarrollando actividades que conducen a la elaboración de información de utilidad tanto para los profesionales que trabajan en los diagnósticos y posibles tratamientos como para las personas afectadas. El Proyecto organiza y financia **2 tipos de actividades de formación**: "**Estancias científicas cortas**" de 5 a 30 días en un centro para establecer o conocer nuevas colaboraciones y "**DSDnet Training School**" en la que profesionales multidisciplinares jóvenes reciben y participan en un programa de formación sobre temas relacionados con DSD: el primero se desarrolló en Ghent (Junio 2015) y el segundo recientemente en Bologna (Octubre 2015).



# A Systematic Elucidation on Differences of Sex Development

BMBS COST Action BM1303

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COST Description
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## Participating Countries

- Austria
- Belgium
- Bulgaria
- Croatia
- Cyprus
- Denmark
- Estonia
- Finland
- France
- Germany
- Hungary
- Israel
- Italy
- Netherlands
- Norway
- Poland
- Romania
- Slovenia
- Spain
- Sweden
- Switzerland
- United Kingdom

## Near Neighbour Countries

- Egypt - National Research Center (NRC)
- Russian Federation - Endocrinology Research Center

## International Partner Countries

- Australia - Murdoch Childrens Research Institute
- Australia - Prince Henry's Institute of Medical Research, Monash Medical Centre
- Australia - The University of Queensland
- Indonesia - Diponegoro University
- Japan - National Centre of Child Health and Development
- Japan - Wakayama Medical University (WMU)
- United States of America - University of Michigan

DSDnet 2014 | Imprint



ESF Provides the COST Office  
through an European  
Commission contract

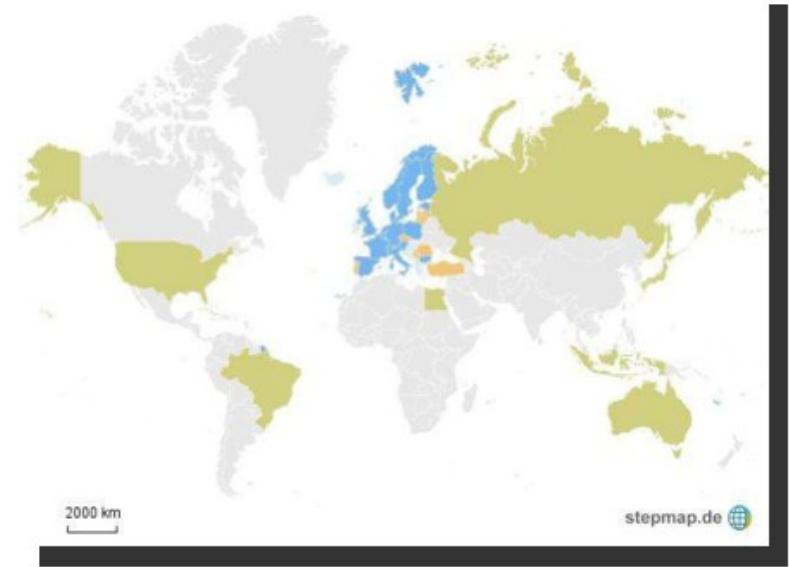
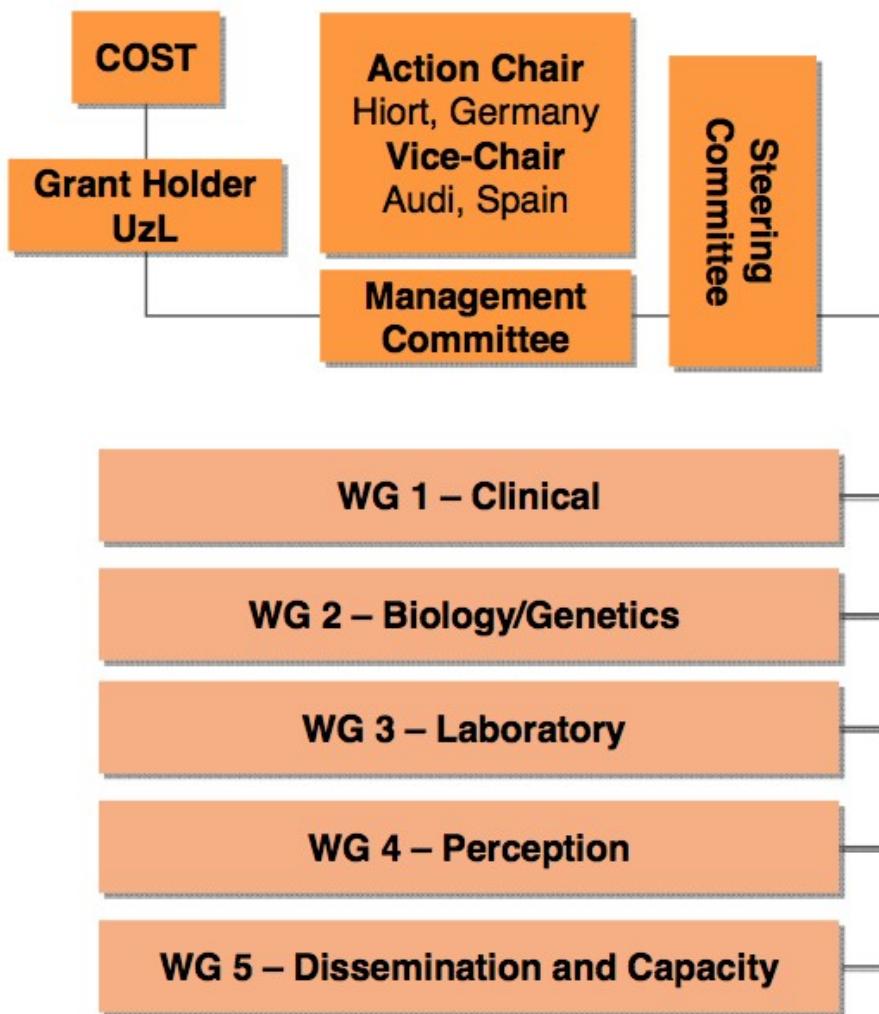


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Programme



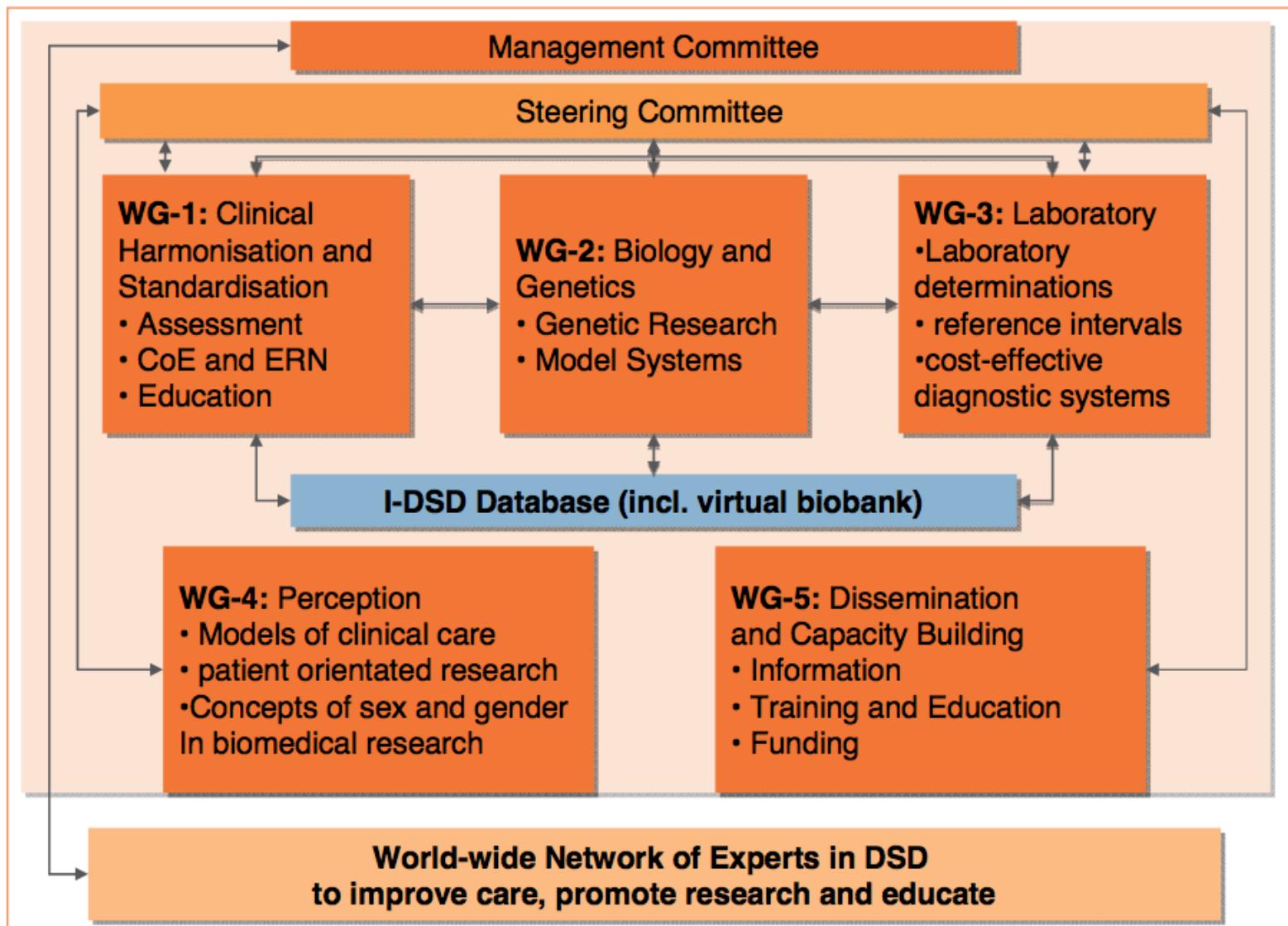
Biomedicine and  
Molecular Biosciences

# Network management, organisation and structure



Connecting all continents  
under European Leadership

# Action Structure





COST is supported by  
the EU RTD Framework  
Programme



ESF Provides the COST Office  
through an European  
Commission contract

Biomedicine and Molecular  
Biosciences





A systematic elucidation of  
Differences of Sex Development  
COST-Action BM1303

# Miembros WG 4:

- **Faisal Ahmed, UK**
- **Arianne Dessens, Netherlands**
- **Jillian Bryce, UK**
- Ira Haraldsen, Norway
- Violeta Iotova, Bulgaria
- **Anders Juul, Denmark\***
- **Maciej Krawczynski, Poland**
- **Andreas Kyriakou, UK**
- Agneta Nordenskjöld, Sweden
- **Caroline Sanders, UK**
- Hedi Van Clahsen\*
- **Marta Rozas, Spain**

# Objetivos del WG 4:

- Explorar modelos de cuidados clínicos y comunicación que faciliten una buena praxis clínica y la investigación.
- Amparar el desarrollo de una protección para grupos de apoyo de personas con DSD.
- Formular las necesidades de los pacientes, orientando la investigación y los futuros juicios clínicos.
- Explorar la aceptación de herramientas de búsqueda de los pacientes.
- Integrar a expertos en bioética y teoría de género.
- Interactuar con los otros WGs para preparar las guías de actuación y las consideraciones enfocadas al paciente.

# WG 4 - Año 2014-

- **Julio 2014:**

- Se comienza el desarrollo de la primera encuesta de expertos de endocrinología pediatrica en centros DSD vinculados a DSDnet y a I-DSD. ✓

- **Septiembre 2014:**

- Prueba de la encuesta con el DSDnet MC (con los directores del proyecto) ✓
- Encuentro en Lubeck

- **Octubre 2014:**

- Se lanza la encuesta a través de los usuarios de DSDnet y I-DSD ✓
- Se comienzan a preparar las encuestas para:
  - pacientes, familiares y grupos de apoyo ✓
  - psicólogos clínicos ✓
  - médicos para adultos, andrólogos, cirujanos

# WG 4 - Año 2015-

- **Febrero 2015:**

- Se lanza la encuesta para los grupos diana – Encuesta para grupos de apoyo a pacientes – Andreas Kyriakou & Marta Rozas – ✓ July 2015
- Se comienza la encuesta para psicólogos clínicos – Arianne Dessens & Guilherme Guaragna – Pendiente de lanzar.

- **Junio 2015:**

- En paralelo a las actividades COST, la universidad de Glasgow organiza un encuentro con un taller de entrenamiento para aprender más de DSD, con médicos, afectados y/o familiares y representantes de grupos. Participan Nuria Grégory Flor y María Martínez Patiño.
- Se estudian los resultados de la encuesta realizada para grupos de apoyo, la cual arroja resultados no deseados.
- Andreas Kyriacou y Marta Rozas presentan en Gante el poster que recoge el trabajo realizado con las encuestas médicas.

# ¿Y con quién se compartió la experiencia de Gante?



**Communication With Peer Support Groups And Families - Results From An International Survey Of Specialist Care For DSD.**  
A. Rajaon A.E. Odeppa J. Ryba I. Haskins V. Rajpa A. Aspi M. Kravczykow A. Narendarid M. Sander O. Hjelg S.F. Ahmed

**Background:** Communication amongst affected people and peer support groups (PSG) is important for optimal management in DSD. However, the extent of communication that occurs at the moment is unclear.

**Methods:** To explore the current models of communication and to determine the current involvement of PSG in DSD care, an international survey of 124 paediatric endocrinologists, identified through DSDNet and the I-DSD Registry, was performed in the last quarter of 2016.

**Results:** A total of 77 (62%) clinicians working in 76 centres and from 22 of 42 countries (51%) responded in 21 (28%) centres; parental individuals with DSD met locally. Of the 77 clinicians, 68 (60%) reported that they are aware of families on PSG and in total, 78 diverse PSG were identified. In 22% of the cases the clinicians reported that they would recommend the affected PSG to the affected person with DSD. Of the 77 clinicians, 27 (35%) reported a collaboration with the MDT during the first three months after a new clinical presentation. In such a case, the availability of a PSG was reported as desirable but not available by 21 (14%) of the clinicians. This group of 27 consisted of 24 (88%) clinicians who were aware of at least one PSG, and 23 (85%) of those not aware of any PSG ( $p=0.003$ ). Discussions of results of genetic tests with the family are lead by a paediatric endocrinologist in 72 (77%) clinics. Other MDT members including a clinical geneticist, in 42 (79%), and clinical psychologist, in 12 (77%) (14%), participate in the discussion with the family. Additional information about the condition was provided to parents by fax or facsimile (9%), direct letter (6%), web-based resources (6%), paper leaflets (2%) and links via PSG (2%). In the communication and information reported to be provided by PSG, 50% used local or national language, 65% in English and 5% in both national and English language.

**Conclusion:** Approximately 50% of paediatric endocrinologists in specialist DSD centres may involve or recommend a formal PSG. There is a need for greater awareness of the availability of local peer support for affected families and with the benefit of this support.

# Los Abstracts

**Diagnostic Approach To A Newborn With Suspected DSD - Results From An International Survey Of Specialist Care For DSD.**

A. Rajaon A.E. Odeppa J. Ryba I. Haskins V. Rajpa A. Aspi M. Kravczykow A. Narendarid M. Sander O. Hjelg S.F. Ahmed

**Background:** The approach to investigating a newborn with a suspected DSD is likely to vary between centres and may be influenced by local availability and technological developments.

**Methods:** To explore the current diagnostic practice and needs, an international survey of 124 paediatric endocrinologists, identified through DSDNet and the I-DSD Registry, was performed in the last quarter of 2016.

**Results:** A total of 77 (62%) clinicians in 76 centres, from 22 of 42 (51%) countries responded to the survey. In a suspected case of 64 (80%) DSD, the investigation that would be performed normally within the first week of presentation included ultrasound (97%), karyotype (95%), ultrasound (94%), 17-OHP (83%), amylase (75%), DHE (75%) XY probe by FISH/PCR (67%), caesarean (67%) and AMH (57%). Second-line investigations included further imaging (80%), array CGH (57%), cDNA (ACTH stimulation) (67%), karyogram (62%) and urinary steroid profile (13%) (1%). The diagnostic tests reported to be not available locally but available included LHRH (1%), array CGH (1%), DHE (1%) and AMH (1%). Clinicians reported that locally they had access to the following genetic tests: SRY (7%), AR (6%), SLC35A2 (2%), NR3C1 (57%), cyp450 genotyping (57%), P450 (7%), DAX1 (5%), SLC35B (5%) and SLC35C (5%) only if family history or biochemical were suggestive. For diagnosing 5-alpha reductase deficiency, 49% clinicians reported genetic testing as the single most preferable test while 25% and 12% reported testosterone/DHE ratio and LH/SH, respectively. The corresponding figures for 17-OHSDS deficiency were 57%, 22% and 12%, respectively.

**Conclusion:** There is considerable variation in the diagnostic evaluation of a newborn with suspected DSD between centres and access to specialist tests may influence this factor. Molecular genetic testing is increasingly common in specialist centres. Clearer guidance in complex cases and collaboration through a network of centres could standardise the need as well as access to diagnostic investigations for DSD.

**Current Models Of Practice & Professional Development Of Clinicians In DSD Centres - Results From An International Survey Of Specialist Care For DSD.**

A. Rajaon A.E. Odeppa J. Ryba I. Haskins V. Rajpa A. Aspi M. Kravczykow A. Narendarid M. Sander O. Hjelg S.F. Ahmed

**Background:** In the optimal care of children with Disorders of Sex Development (DSD), it is generally considered good practice to work within a multidisciplinary team (MDT) and engage in opportunities for professional development.

**Methods:** To explore the current models of MDT practice and the extent of professional development in specialist DSD centres, an international survey of 124 paediatric endocrinologists, identified through DSDNet and the I-DSD Registry, was performed in the last quarter of 2016.

**Results:** A total of 77 (62%) clinicians in 76 centres, from 22 of 42 (51%) countries responded to the survey. In 61 (81%) of the centres, the lead of the team that provided DSD care was a paediatric endocrinologist with the next commonest being a clinical geneticist in 5 (7%) centres. The surveyed clinicians responded that the following paediatric specialists would be routinely involved in the initial evaluation of a newborn - endocrinologist (95%), surgeon/urologist (91%), radiologist (90%), neonatologist (90%), clinical geneticist (81%) and clinical psychologist (81%). However, during the first week after presentation, a team consisting of paediatric specialists in endocrinology, surgery/urology, clinical psychology, neonatology and nursing was only possible in 29 (76%) of centres. Over the first three months after presentation, a team comprising of paediatric specialists in endocrinology, surgery/urology, clinical psychology, nursing and clinical genetics was only possible in 11 (24%) of the centres. A nationally organised network or plan for managing rare conditions such as DSD was reported only in 10 (13%) (7%) centres. Of the 77 clinicians, 23 (30%) kept a local DSD registry only, 40 (52%) shared their data in a multicentre DSD registry and 9 (12%) did not record any data. Participation in audit/quality improvement exercises in DSD care was reported by 12 (16%) centres in 6 (21%) (5%) countries. Attendance in local, national or international DSD related educational programs was reported by 60% (39%) and 82% (62%) clinicians, respectively. Case discussions (30%) and conferences/training days (5%) provide the main opportunity for centres to improve the knowledge in DSD of health care professionals outside their own centre.

**Conclusion:** Although an increasing number of DSD centres have access to paediatric staff, the actual delivery and quality of care provided by these staff requires further exploration. Professional development and engagement in activities that may lead to improved care need further attention.

# Los Poster



Symposium Day 2 (Friday 12/06/2015)

## Poster Presentations

Gallery, New Zebra

Posters will be displayed on Friday 12<sup>th</sup> June in the Gallery. Posters should be mounted in the morning and removed after the end of the attended poster session (4-5pm).

- P1 Communication With Peer Support Groups And Families - Results From An International Survey Of Specialist Care For DSD. Marta Rocas, Support Group GrApSA, Barcelona, Spain
- P2 Follow-Up Studies: The Good, The Bad, and The Ugly. Michael Kreutzer, University of Ottawa, Canada
- P3 Sex Testing of Elite Female Athletes with DSD: Science and Controversies. María José Martínez-Patiño, University of Vigo-Institute of Biomedical Research (IB-Uvigo), Spain
- P4 Incorporating Support & Advocacy into the Disorders of Sex Development – Translation | Research Network (DSD-TRN): Building Relationships and Resources for Patients, Families, and Healthcare Providers. Janet Green, Accord Alliance/DSD-TRN
- P5 Applying Hume's Is/Ought Problem to 'Disorder of Sex Development'. Natalie Delimat, Institute of Technology Sligo, Ireland.
- P6 Body Image and Quality of Life (QoL) in women with Congenital Adrenal Hyperplasia - outcomes and avenues for adjuvant treatment . Erika Tomlinson, Austin/Northern Health, Melbourne, Australia
- P7 Psychosocial well-being in Dutch adults with a disorder of sex development. N.G.M. de Neve-Enthoven, Erasmus Medical Center Rotterdam, Netherlands
- P8 Psychological consequences for DSD patients with Ychromosome. Katarzyna Bajszczak, Children's Memorial Health Institute, Warsaw, Poland
- P9 Health care-seeking behavior in late-identified patients with congenital adrenal hyperplasia (CAH) in Central Java, Indonesia. Agustini Utari, Diponegoro University, Indonesia
- P10 The body image and genitalia appearance concerns and virilization features of CAH female patients in Malaysia. Ani Amelia Zainuddin, The National University of Malaysia (UKM), Kuala Lumpur, Malaysia
- P11 Islamic perspectives of DSD and gender-related issues. Ani Amelia Zainuddin, The National University of Malaysia (UKM)
- P12 "We are also humans we who are sick" Experiences of 13 women with congenital adrenal hyperplasia . Hedvig Engberg, Karolinska Institutet and Child and Adolescent Psychiatry Research Center, Stockholm, Sweden
- P13 Multidisciplinary and psychosocial shift in DSD/Intersex management. Nuria Gregori Flor, Valencia University, Spain
- P14 Three novel CYP21A2 mutations identified in Brazilian patients with 21-Hydroxylase deficiency: A synergistic effect. Débora de Paula Micheletto, Karolinska University Hospital, Sweden
- P15 FGFR2 mutation in XY sex reversal with craniostenosis . Vincent Harley, MIMR-PHI Institute of Medical Research, Melbourne, Australia



# Communication With Peer Support Groups And Families

## - Results From An International Survey Of Specialist Care For DSD



J. Kyratzi<sup>1</sup>, M. Rønne<sup>2</sup>, A.B. Deesche<sup>3</sup>, L. Bryce<sup>4</sup>, L. Haraldsen<sup>5</sup>, V. Iotova<sup>6</sup>, A. Jutly<sup>7</sup>, M. Krawczynski<sup>8</sup>,  
A. Nordendijk<sup>9,10</sup>, C. Sanders<sup>11</sup>, O. Hirtz<sup>12</sup>, S.F. Ahmed<sup>13</sup>

<sup>1</sup>. University of Glasgow, UK <sup>2</sup>. Great Ormond Street Hospital, MC-Baskin Children's Hospital, Amsterdam, The Netherlands <sup>3</sup>. Oslo University Hospital, Norway <sup>4</sup>. Medical University of Vienna, Austria <sup>5</sup>. University of Copenhagen, Denmark <sup>6</sup>. Paris University of Medical Sciences, France <sup>7</sup>. Karolinska University Hospital, Stockholm, Sweden <sup>8</sup>. Alder Hey Children's Hospital, UK <sup>9</sup>. University of Ulm, Germany

### Introduction - Aim

Communication amongst affected people and support groups (PSG) is important for optimal management of DSD. However, the extent of communication that occurs at the moment is unclear.

The aim of the study was to explore the current models of communication between clinicians and affected people and to determine the current available forms of communication groups in DSD care.

### Methods

An international survey of centres that deliver specialist care for children with DSD was performed from 1st October 2014 to 7th December 2014.

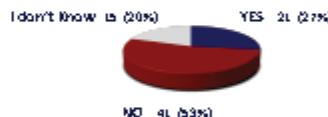
124 clinicians working in the field of paediatric endocrinology (selected through the DSDnet and the I-DSD Registry) were invited to participate in the survey.

### Results

#### Response Rate

Region	Type		Response rate		Number of responses	Number of children
	Child	Adult	Child	Adult		
Europe	8%	11%	11%	10%	14	41
America	7%	11%	11%	11%	11	33
Asia & Africa	5%	4%	4%	4%	4	12
Africa	0%	0%	0%	0%	0	0
Oceania & Middle East	0%	0%	0%	0%	0	0
Total	4%	6%	20%	16%	34	100

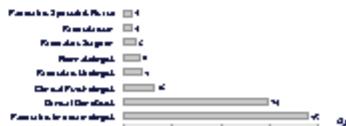
Do parents/individuals with DSD in your centre meet locally?



How is information about the condition provided to the family?



Who discloses the results of the genetics with the family?



### Conclusion

As a result of a lack of standardised disclosure, especially DSD clinics may have a recommendation to form local support groups. This is a need for greater awareness of the availability of local support groups for affected families, as well as the benefits of the support.

# Poster Andreas K. & Marta R.

#### DSDNet: Communication With Peer Support Groups And Families - Results From An International Survey Of Specialist Care For DSD

4. *Hydrogen*? 5. *Boron*? 6. *Iron*? 7. *Neutron*? 8. *Proton*? 9. *Antineutron*?  
10. *Antiproton*? 11. *Positron*? 12. *Muon*? 13. *Electron*?

3. Measures of Disease Off-Label Use: A review of prescription drug use in the United States. Washington, DC: American Progress; 2010. Available online at [www.americanprogress.org/issues/2010/07/reports/10000/report.pdf](http://www.americanprogress.org/issues/2010/07/reports/10000/report.pdf). Accessed 17 October 2010.

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...and the first time you see your new home, you'll know it's a real "home". It's built to last a lifetime.

10. The following table shows the number of hours worked by 1000 workers in a certain industry.

10

For more information about the program, contact the Office of the Vice Provost for Research at 216-368-2300 or visit [www.case.edu/research/](http://www.case.edu/research/).

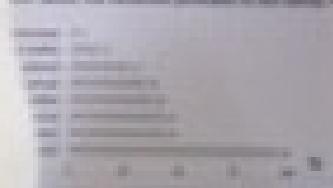
and more recently in the field of quantum optics,<sup>1</sup> through the famous work of Schrödinger, Heisenberg, and Dirac.

100

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- **Agosto 2015:**

- Se debería completar la inclusión de detalles de centros DSD en la DSDnet - Pendiente de discutir con el WG 5 los detalles de los datos.

- **September 2015:**

- Se celebra el segundo encuentro de los WG y el tercero del los MC en Poznan:
    - Presentación del informe de resultados de las encuestas ✓ Detalle de los miembros de DSDnet
    - Workshop – Enfocado en grupos de apoyo
    - Presentación en los encuentros internacionales ✓

# Planes futuros

## Completar para finales de 2015:

- ✓ Inclusión de detalles de los centros en DSDnet
- ✓ Borrador de encuesta de pacientes y padres en las clínicas: Caroline, Joanne, Andreas & Marta. Actualmente se están celebrando reuniones a través de Skype para trabajar en la encuesta fallida de grupos de apoyo.
- ✓ Incluir científicos sociales en el WG4- Caroline Sanders
- ✓ Considerar encuestas para otros profesionales- ginecólogos, cirujanos.
- ✓ Cooperar con WG2 y WG3, qué información precisan?
- ✓ Enviar un manuscrito de encuesta endocrino pediátrica
- ✓ Encuesta para psicólogos

# Y para 2016

## **Primavera 2016**

- Videoconferencia – WG4
- Encuesta de pacientes completada

## **Mediados 2016:**

- Workshop para pacientes y padres. El WG 4 ya está trabajando en ello.
- Explorar la aceptación para pacientes y usuarios de las herramientas médicas y de búsqueda.
- Determinar las necesidades de los pacientes – búsqueda orientada y ensayos clínicos.

## **Sept 2016:**

- Tercer encuentro de los WG
- Revisión

## **Finales 2016**

- Cambiar la composición de los WG- Expertos en bioética y teoría de género.

## **Comienzos 2017**

- Repetir las encuestas iniciales de 2014/2015