

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

[HOME](#) | [DSDNET](#) | [INFORMATION](#) | [EDUCATION](#) | [NEWS/ EVENTS](#) | [RESEARCH](#) | [I-DSD](#) | [CONTACT](#)

[DSDnet](#) > Parties

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

[COST Description](#)

[Parties](#)

[Working Groups](#)

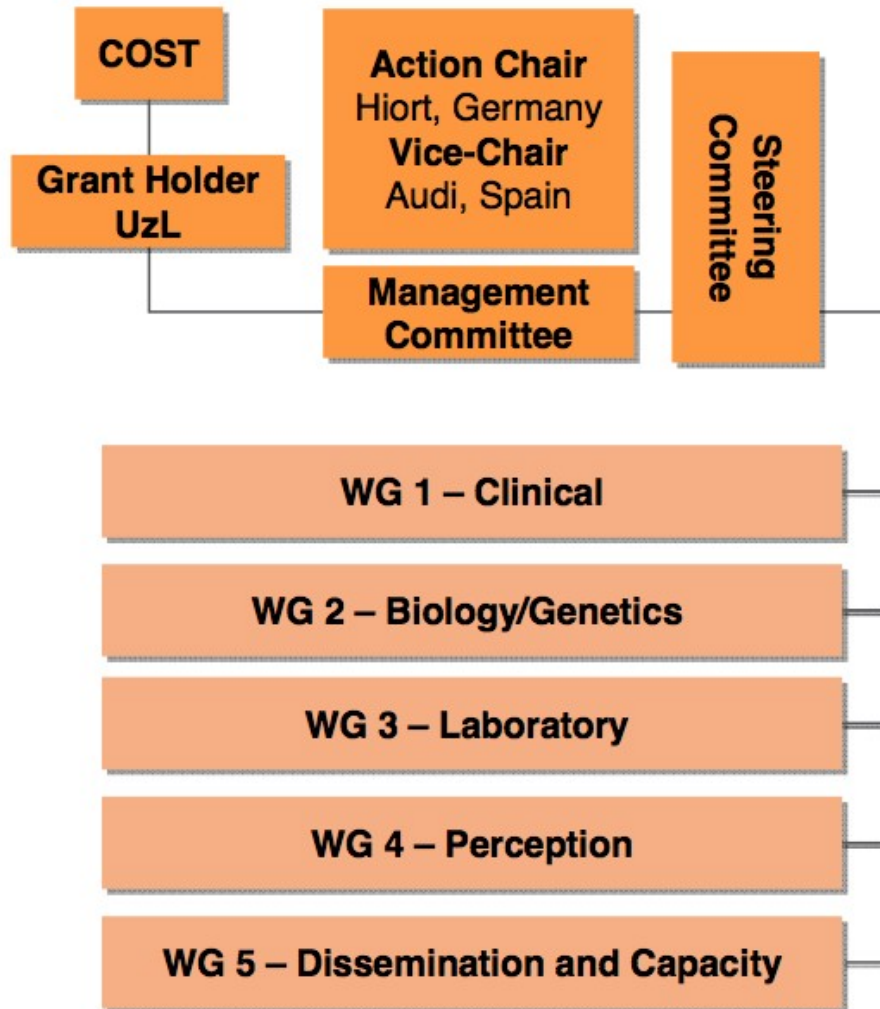
[Management Committee](#)

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[Profil](#)
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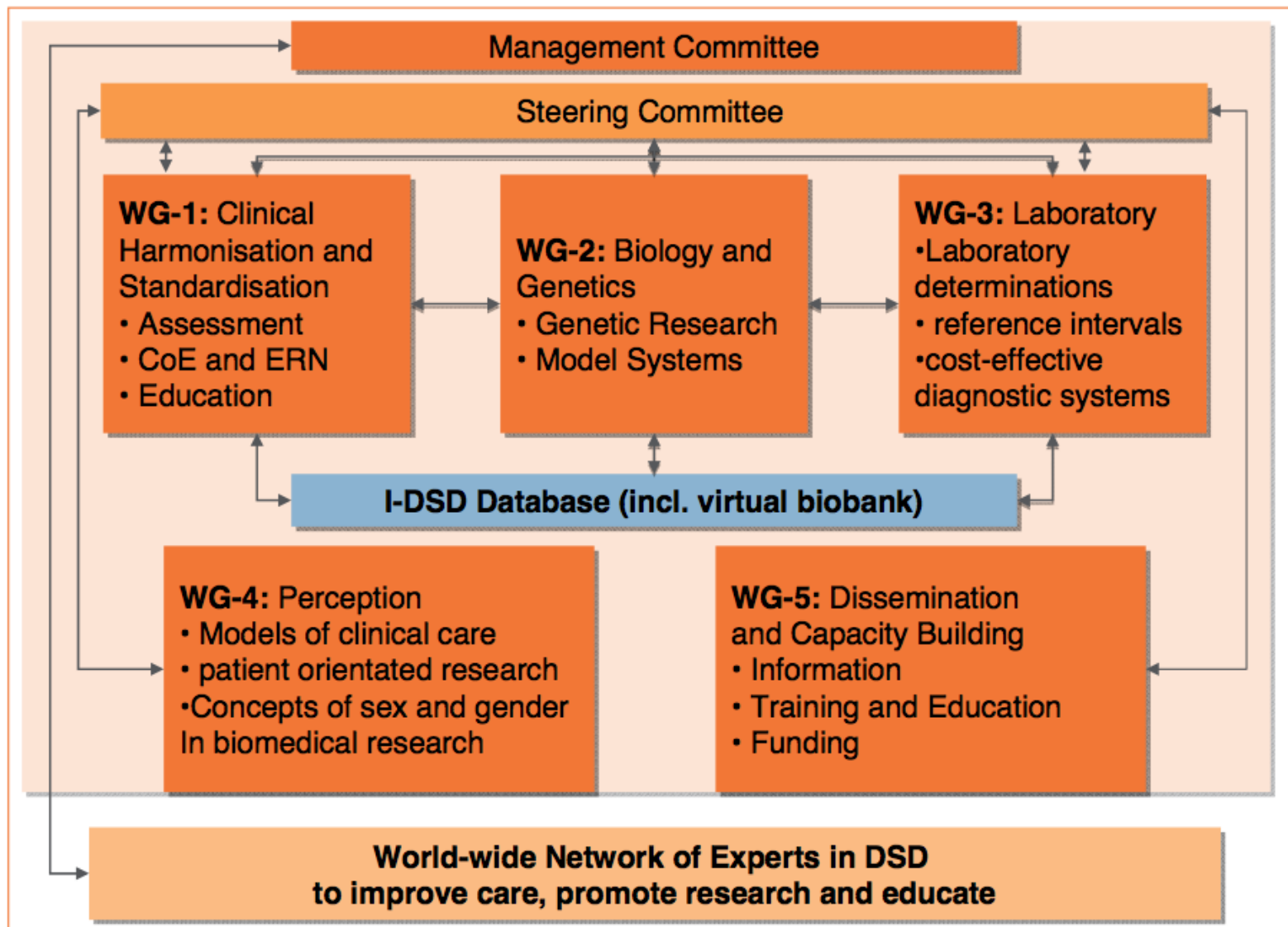
DSDnet 2014 | Imprint

Network management, organisation and structure



Connecting all continents
under European Leadership

Action Structure





ESF Provides the COST Office
through an European
Commission contract



COST is supported by
the EU RTD Framework
Programme

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 Claahsen*
- 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 pediátrica 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?



Los Abstracts

Communication With Peer Support Groups And Families - Results From An International Survey Of Speech/Life Care For ASD

A. Kuznetsov, M. Raza, A.B. Qasbi, I. Bryce, I. Hamidov, V. Sazon, A. Jari, M. Kuznetsov, A. Nordin, M. C. Santos, O. Elom, S.E. Ahmed

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

Methods: To explore the current mode of communication and to document the current involvement of PSC in DSD care, an international survey of 138 pediatric endocrinologists, identified through [GGOpp](#), and the [I-DSD Registry](#), was performed in the last quarter of 2016.

Results: A total of 70/138 (50%) clinicians working in 74 centers and from 28 of 62 countries (45%) responded in 21 (28%) centers, parented individuals with DSD were locally

Of the 77 clinicians, 69 (90%) reported that they are aware of the clinical usage and, instead, 76 clinicians (99%) were identified. In 82% of the cases the clinicians reported that they would recommend the spend upon the affected person with DSD. Of the 77 clinicians, 73 (95%), reported a collaboration with the MDT during the first three months after a new clinical presentation. In such a scenario, the availability of a PGG was regarded as desirable but not available by 61 (81%) of the clinicians. This group of 61 consisted of 26 (43%) any clinician who were aware of it, 14 (23%) PGG and 21 (35%) PGG; of those not aware of any PGG ($n=16$) (21%). Discussions of the results of genetic tests with the family are led by a paediatric endocrinologist in 74 (77%) of the clinicians. Other MDT members including a clinical geneticist, in 52 (73%) and clinical psychologist, in 12 (17%) (14%) participate in the discussion with the family. Additional information about the condition was provided to patients by a nurse in 34 (discretion/PGG), clinical geneticist (66%), web-based resource (42%), parent (infant) (31%) and links with PGG (23%). In the communication and information provided to be provided by PGG, 55% via text local or national language, 45% in English and 4% in both regional and Spanish language.

Conclusion: Approximately 50% of pediatric endocrinologists in specialist DSD centers may involve or recommend a formal FSG. There is a need for greater awareness of the availability of local resources for affected families as well as the benefit of this support.

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

A. Kuznetsov, A. B. Gerasimov, I. Bryuk, I. Hladchenko, V. Isakov, A. Jap, M. Kravtsov, A. Nondendakid, M. Raza, C. Sander, O. Shv, S. E. Ahmed

Background: The approach to investigating a newborn with a suspected OSD 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 120 paediatric endocrinologists, identified through [ORCID](#) and the I-CED Registry, was performed in the last quarter of 2016.

Results: A total of 1102/6724 chondroblasts in 76 cases, from 26/32/29/34 countries, registered to the survey. In a subgroup of 63/403/252, the investigation of the mutation was performed routinely within the first week of presentation (individuals: 59/34/19; ages: 40/66/39); ultrasound/9/34; 17-GAG/17/34; and ALP/25/34). DMS (7/34) was probed by FISH (3/34); *COL2A3* (1/34) and *COL2A3/25*. Second-line investigations included further imaging/3/34; array CGH/3/34; *COL2A3* ACTH mutation/3/34; *COL2A3* duplication/2/34; and urinary steroid profile/1/34/34. The diagnostic test reported as not available locally but detectable included USP/3/34; array CGH/1/34; DMS/1/34; and ALP/1/34. Chondroblasts exposed that locally they had access to the following genetic tests: USP/1/34; ALP/3/34; DMS/3/34; *N/25/34*; *COL2A3* genomic analysis/5/34; 17/7/34; *COL2A3*; *COL2A3*; and a wider range of genes/6/34. The genetic tests on chondroblasts would perform routinely in a case of X-Linked DSD included USP/3/34; ALP/1/34; *N/25/34* and *N/25/34/25*, while they would perform: *COL2A3*/1/34; 17/7/34; *N/25/34*; *COL2A3*; *COL2A3/25/34* and *COL2A3/25/34* only if family history or biochemical tests suggested. For diagnosing to molecular defects, 6/34 chondroblasts reported genetic testing at the single most profitable time while 3/34 and 1/34 reported *COL2A3* mutation into USP, respectively. The corresponding figures for 17GAGH deficiency were 5/34, 2/34 and 1/34. 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 paediatric cases. Clear guidance on coordination and collaboration through a network of centres could anticipate 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 Speechlet Care For DSD.

A. Katsalou, A. M. Gossens, I. Bryce, I. Haidich, V. Lopez, A. Japi, M. Krawczynski, A. Nonduradillo, M. Raza, C. Sander, O. Hart, S. E. 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 ensure an environment 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 126 paediatric endocrinologists, identified through [Q66type](#) and the I-DSD Registry, was performed in the last quarter of 2014.

Results: A survey of 1126 (62%) clinicians in 74 countries, from 28 (17%) countries responded to the survey. In 61(24%) of countries the clinical team that provided DSD care was a pediatric endocrinologist with the next commonest being clinical geneticist in 5(7%) countries. The surveyed clinicians reported that the following pediatric specialties would be routinely involved in the initial evaluation of a newborn - endocrinology(93%), surgery/oncology(92%), radiology(74%), neurology(50%) clinical genetics(41%) and clinical psychology(69%). However during the first week after presentation, a team concerning of pediatric endocrinology in endocrinology, surgery/urology, clinical psychology, neurology and nursing was only possible in 29(70%) of countries. Over the first three months after presentation, a team comprising of pediatric endocrinology, surgery/urology, clinical psychology, nursing and clinical genetics was only possible in 31(74/61%) of the countries. A nationally organized network or plan for managing rare conditions such as DSD was reported to exist in 14(56/74) countries. Of the 74 clinicians, 28(34%) kept a log of DSD registry only, 40(51%) shared their data in a multicentre DSD registry and 6(7%) did not record any data. Participation in audit quality improvement courses in DSD was not reported by 13(16/74) countries, 6(8/15%) countries. Attendance in local, national or international DSD related educational programs was reported by 69% (74), 32% and 22% clinicians, respectively. Case discussions(30%) and conferences/training days(34%) provided the main opportunity for countries to improve the knowledge in DSD of health care professionals outside their own country.

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

Los Poster

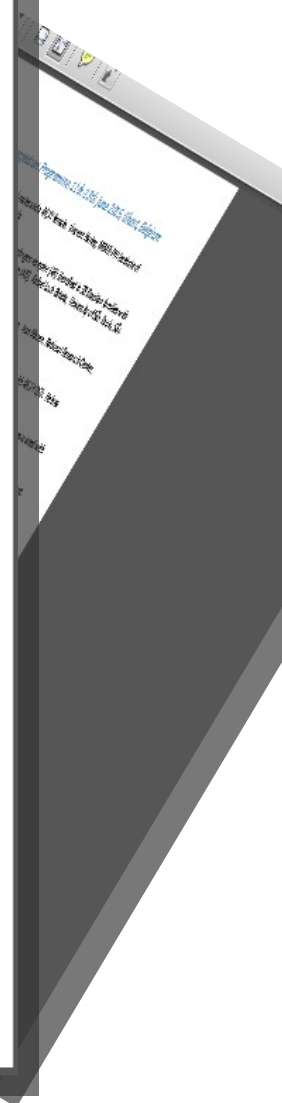
Symposium Day 2 (Friday 12/06/2015)

Poster Presentations

Gallery, New Zebra

Posters will be displayed on Friday 12th 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. Maria Rozas, Support Group GrAPdIA, Barcelona, Spain
- P2 Follow-Up Studies: The Good, The Bad, and The Ugly. Michael Kreuzer, University of Ottawa, Canada
- P3 Sex Testing of Elite Female Athletes with DSD: Science and Controversies. Maria José Martínez-Ratiño, University of Vigo-Institute of Biomedical Research (IB-Vigo), Spain
- P4 Incorporating Support & Advocacy into the Disorders of Sex Development - Translational 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 Delmar, 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 Nijve-Enthoven, Erasmus Medical Center Rotterdam, Netherlands
- P8 Psychological consequences for DSD patients with Y-chromosome. Katarzyna Bajczak, Children's Memorial Health Institute, Warsaw, Poland
- P9 Healthcare-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 Za'inuddin, The National University of Malaysia (UKM), Kuala Lumpur, Malaysia
- P11 Islamic perspectives of DSD and gender-related issues. Ani Amelia Za'inuddin, 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 Michelatto, Karolinska University Hospital, Sweden
- P15 FGFR2 mutation in XY sex reversal with craniosynostosis. Vincent Hailey, 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

K. Kyriakou¹, M. Ross², A.B. Dossena³, L. Blythe⁴, L. Haraldsson⁵, V. Kotova⁶, A. Juul⁷, M. Krawczynski⁸,
A. Nordenskjöld⁹, C. Sanders¹⁰, O. Hørt¹¹, S.F. Ahmed¹²

1. University of Glasgow, UK; 2. GUSIS, 3. S. H. M. M. MC-Schake Children's Hospital, Rotterdam, The Netherlands; 4. Oslo University Hospital; Norway; 5. Medical University of Vienna, Bulgaria; 6. University of Cologne, Germany; 7. Poznań University of Medical Sciences, Poland; 8. Caroluska University Hospital, Stockholm, Sweden; 9. Ålderley Children's Hospital, UK; 10. University of Ulster, Germany

Introduction - Aim

Communication amongst affected people and peer 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 role of peer support groups in DSD care.

Methods

An international survey of centres that deliver specialist care for children with DSD was performed from 15th October 2014 to 2nd December 2014.

124 clinicians working in the field of paediatric endocrinology, referred through the DSDnet and the I-DSD registry, were invited to participate in the survey.

Results

Response Rate

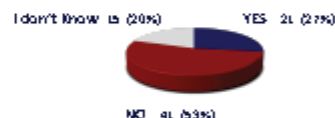
Region	All centres		Children's		General		Specialist	
	n	%	n	%	n	%	n	%
Europe	62	51	47	76	36	58	47	76
North America	7	6	5	71	2	33	2	33
South America	5	4	4	80	1	20	4	80
Africa	7	6	6	86	1	14	1	14
Asia and Oceania	11	9	10	91	1	9	1	9
Total	92	76	72	78	40	43	54	59

Communication with Peer Support Groups

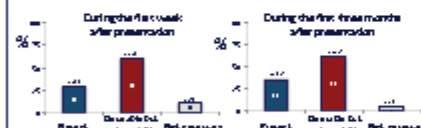
Willingness to recommend the reported group to affected person/parents



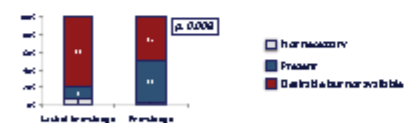
Do parents/individuals with DSD in your own region locally?



Collaboration of Support Groups with the multidisciplinary team

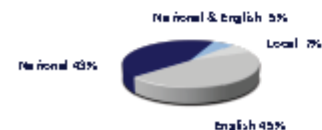


Of the 77 children, 48 (62%) reported that they were aware of at least one PSG



A PSG was reported as desirable but not available from 50% of the children who were aware of at least one peer support group, and from 75% of those not aware of any PSG.

Language in which the information is provided by Support Groups



Conclusion

As a consequence of paediatric endocrinology, specialist DSD centres may have been recommended a formal peer support group. There is a need for greater awareness of the availability of local peer support for affected families as well as the benefits of the support.

Poster
Andreas
K.
&
Marta R.

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

A. Kumbak¹, M. Beale², A.B. Brown³, J. Brown⁴, J. Hordburn⁵, A. Jones⁶, A. Joffe⁷, M. Kumbak⁸,
 A. Kumbak⁹, J. Kumbak¹⁰, G. Kumbak¹¹, L.J. Kumbak¹²

¹ University of Glasgow, UK; ² University of Glasgow, UK; ³ University of Glasgow, UK; ⁴ University of Glasgow, UK; ⁵ University of Glasgow, UK; ⁶ University of Glasgow, UK; ⁷ University of Glasgow, UK; ⁸ University of Glasgow, UK; ⁹ University of Glasgow, UK; ¹⁰ University of Glasgow, UK; ¹¹ University of Glasgow, UK; ¹² University of Glasgow, UK

Introduction

Communication with peer support groups and families is an important part of specialist care for DSD. However, the extent of communication with these groups is unclear. This study aims to explore the current practice of communication with peer support groups and families in specialist care for DSD.

Methods

An international survey of specialist care for DSD was conducted from 1st October 2012 to 31st December 2012. All specialist working in the field of specialist care for DSD through the DSD and the DSD register were invited to participate.

Results

Response Rate

Country	Invited	Response	Response Rate (%)
UK	10	8	80
USA	5	4	80
Canada	3	2	67
Australia	2	1	50
Other	10	7	70
Total	30	22	73

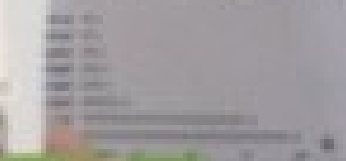
For parents/families with children or young people with DSD



How often the specialist provided to the family



Methods of the specialist with the family



Communication with Peer Support Group

Availability of communication for support group to different specialist



Availability of Support Groups with the specialist group



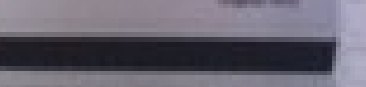
What is the most common method of communication



What is the most common method of communication



Language in which the information is provided to the family



- **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 de 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