Modelos actuales de atención y futuras direcciones en las DSD

Madrid, marzo de 2019

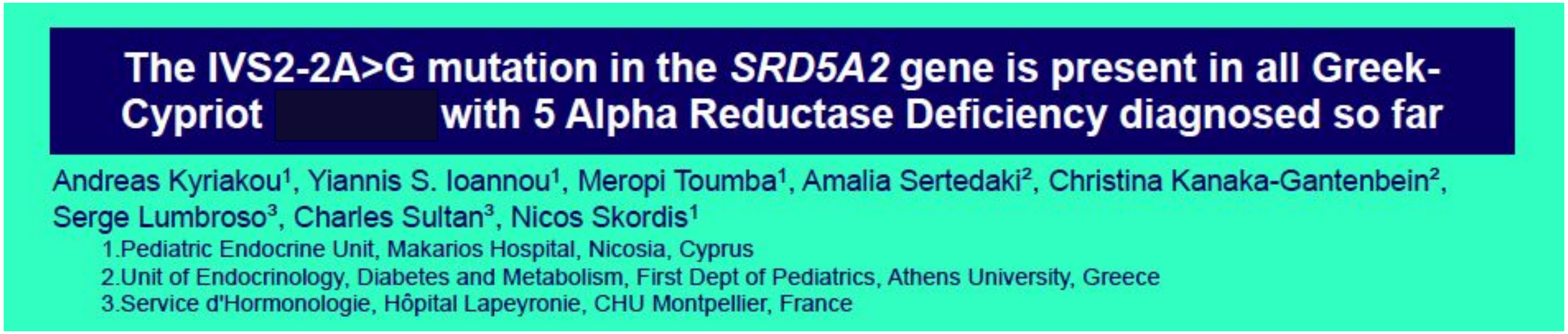
XVIII Encuentro GrApSIA

Dr Andreas Kyriakou

Paediatric Endocrinologist, Glasgow, UK

Year 2006

Presenting 4 cases of a rare DSD



The IVS2-2A>G mutation in the *SRD5A2* gene is present in all Greek-Cypriot [redacted] with 5 Alpha Reductase Deficiency diagnosed so far

Andreas Kyriakou[1], Yiannis S. Ioannou[1], Meropi Toumba[1], Amalia Sertedaki[2], Christina Kanaka-Gantenbein[2], Serge Lumbroso[3], Charles Sultan[3], Nicos Skordis[1]

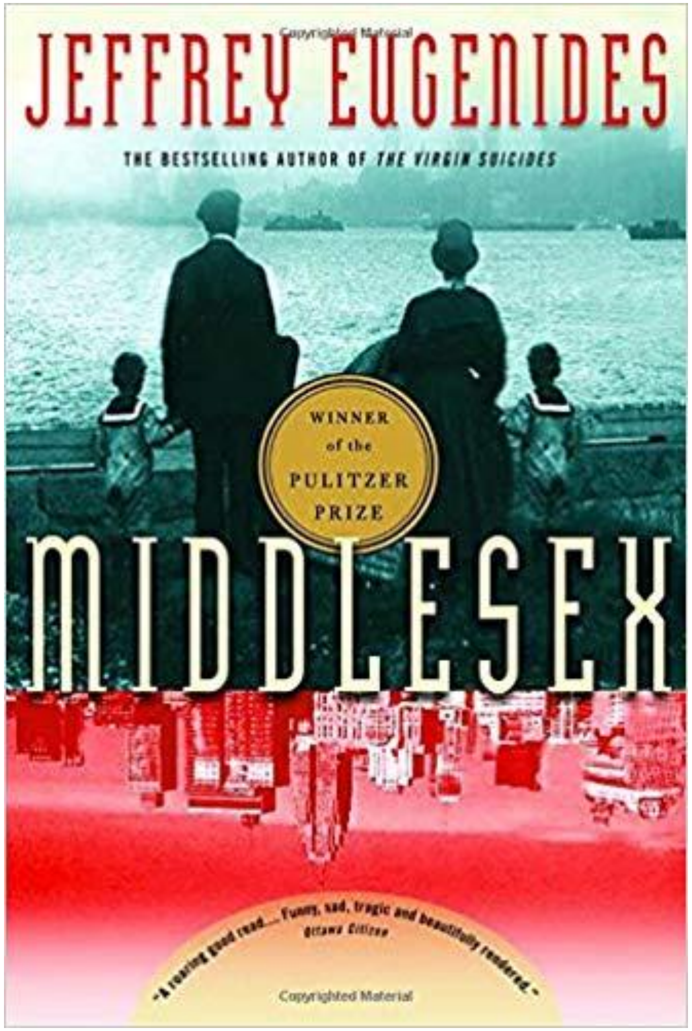
1.Pediatric Endocrine Unit, Makarios Hospital, Nicosia, Cyprus
2.Unit of Endocrinology, Diabetes and Metabolism, First Dept of Pediatrics, Athens University, Greece
3.Service d'Hormonologie, Hôpital Lapeyronie, CHU Montpellier, France

Four girls with a rare DSD condition

- The changes in their body
- Their hormone levels in their body
- The differences in their genes

I was not aware of

- the challenges they may face in their lives
- they were not informed about their condition
- the irreversible surgeries they had without being completely informed



Middlesex

Pulitzer prize-winning novel

- The main character is a person with the same rare DSD

- a fascinating character
- the relationships in adolescence and adulthood
- the person’s emotions and challenges in life

No discussion about genes or doctors or tests

Year 2006

Presenting 4 cases of a rare DSD

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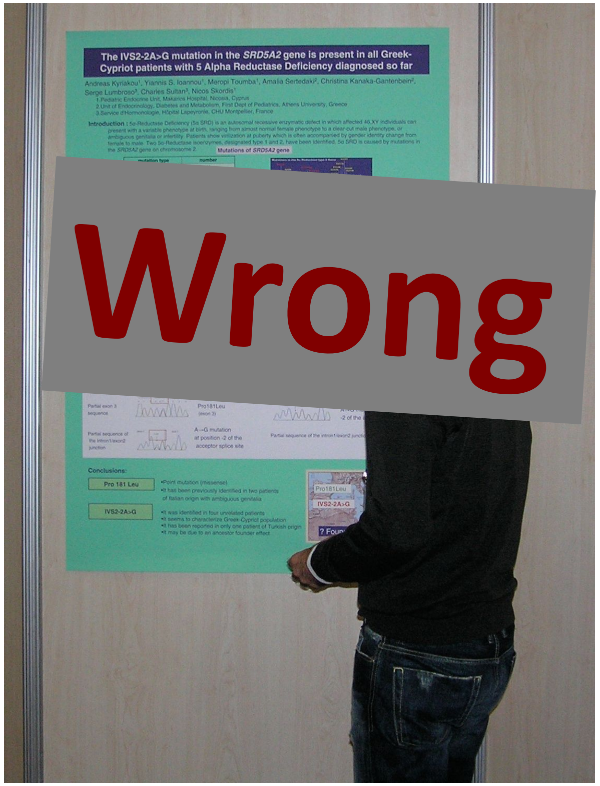


Optimal Care for DSD

Holistic approach

care for all aspects of the life of the person

Life-long

from birth to adulthood

Requirements

- Multidisciplinary care: a wide range of specialists involved
- Collaboration between centres and between countries – to create national and international networks
- Collaboration with families and Support Groups for DSD

International Collaboration

COST Action DSDnet 2013 - 2017

1. **European countries with various cultural and financial background**

   23 European countries
   (with 3 near neighbour and 5 international countries)

2. **Wide range of Specialties**

- Paediatric Endocrinologists
- Psychologists
- Geneticists
- Biochemists
- Support Group members
- Gynaecologists
- Nurses
- Surgeons/Urologists

Working Group

Team: Members from 9 European countries

4 Paediatric Endocrinologists	1 Nurse
2 Psychologists	1 Geneticist
2 Support Group representatives	1 Surgeon

Aiming to explore

1. The current models of clinical care - how do specialist centres work
2. Communication of healthcare professionals with individuals/families and Support Groups
3. The needs for future research

End of Action meeting in February 2018

Current Models Of Care for DSD – Results From An International Survey Of Specialist Centres

A. Kyriakou[1], A.B. Dessens[2], J. Bryce[1], V. Iotova[3], A. Juul[4], M. Krawczynski[5], A. Nordenskjöld[6], M. Rozas[7], C. Sanders[8], O. Hiort[9], S.F. Ahmed[1]

1. Glasgow, UK 2. Rotterdam, The Netherlands 3. Varna, Bulgaria 4. Copenhagen, Denmark 5. Poznan, Poland 6. Stockholm, Sweden 7. GrApSIA Spain 8. Liverpool, UK 9. Lübeck, Germany

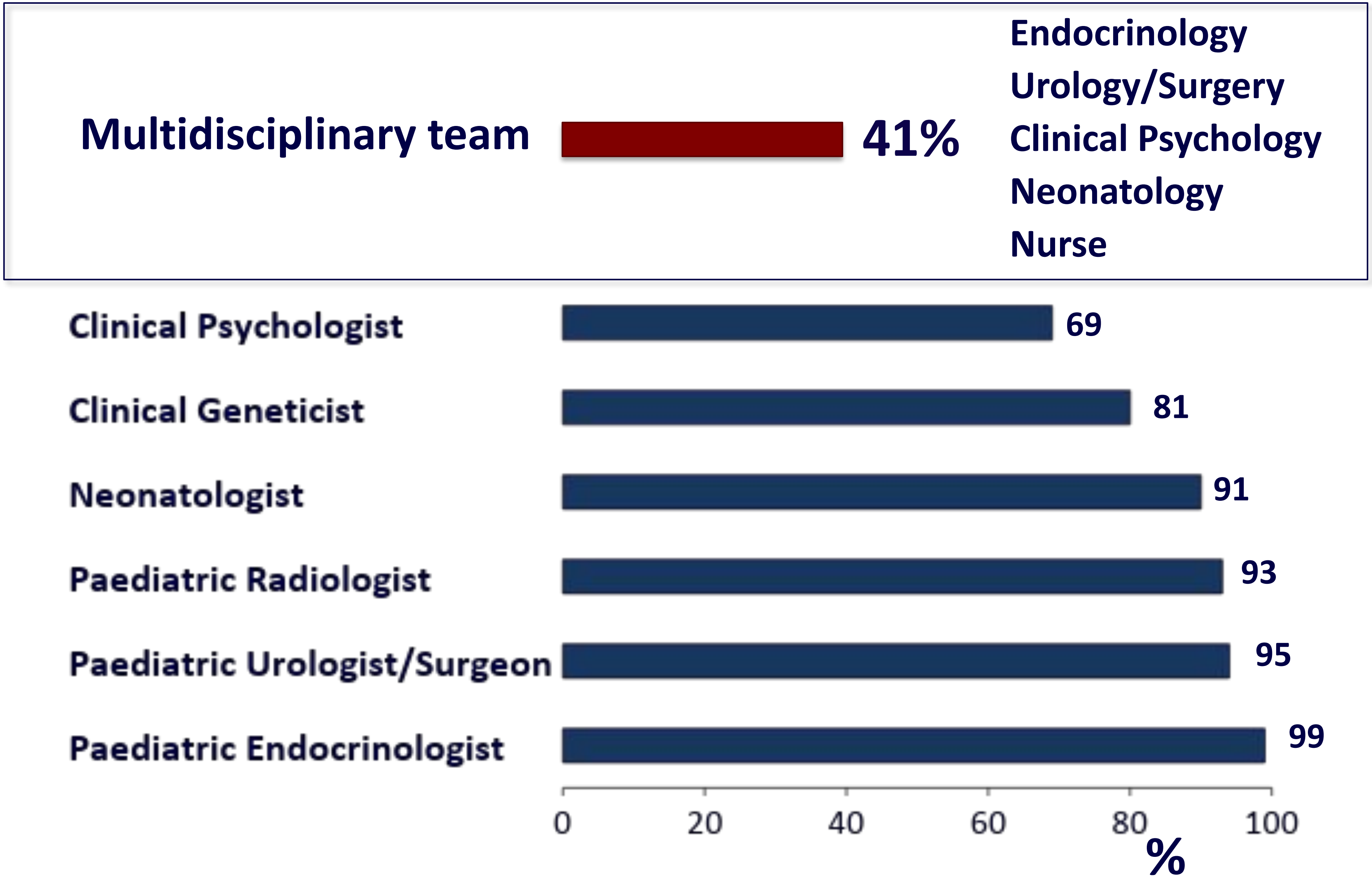
Kyriakou et al. Orphanet Journal of Rare Diseases (2016) 11:155

International Survey of Clinicians/Centres delivering Specialist Care for DSD

Questionnaire about the current clinical practice

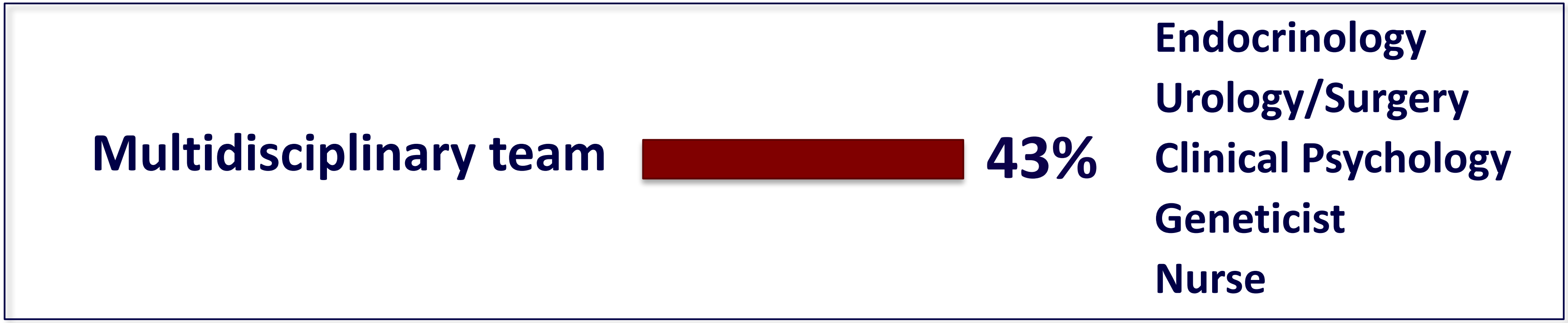
Response	Doctors	Centres	Countries
	78	75	38

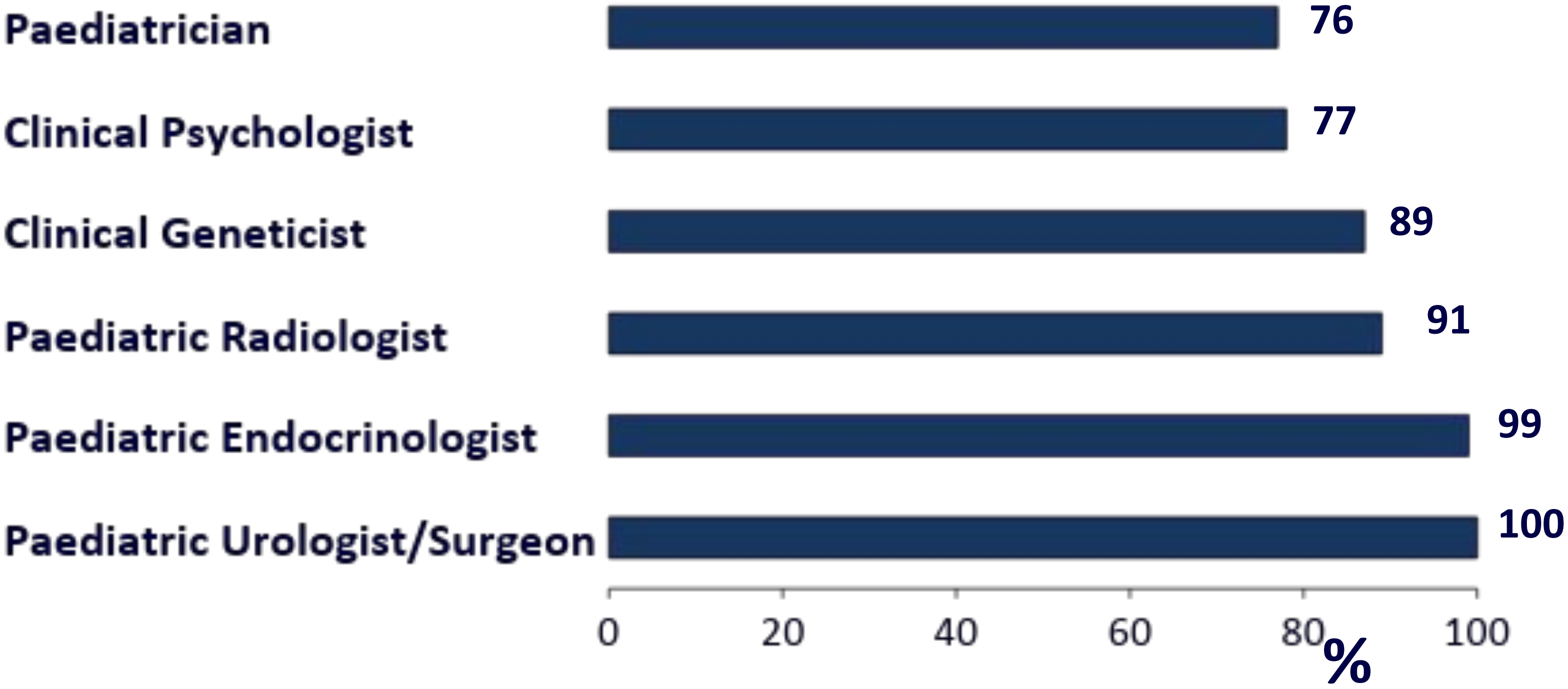
Specialists involved in the initial evaluation of a newborn with DSD - At diagnosis



Endocrinology
Urology/Surgery
Clinical Psychology
Neonatology
Nurse

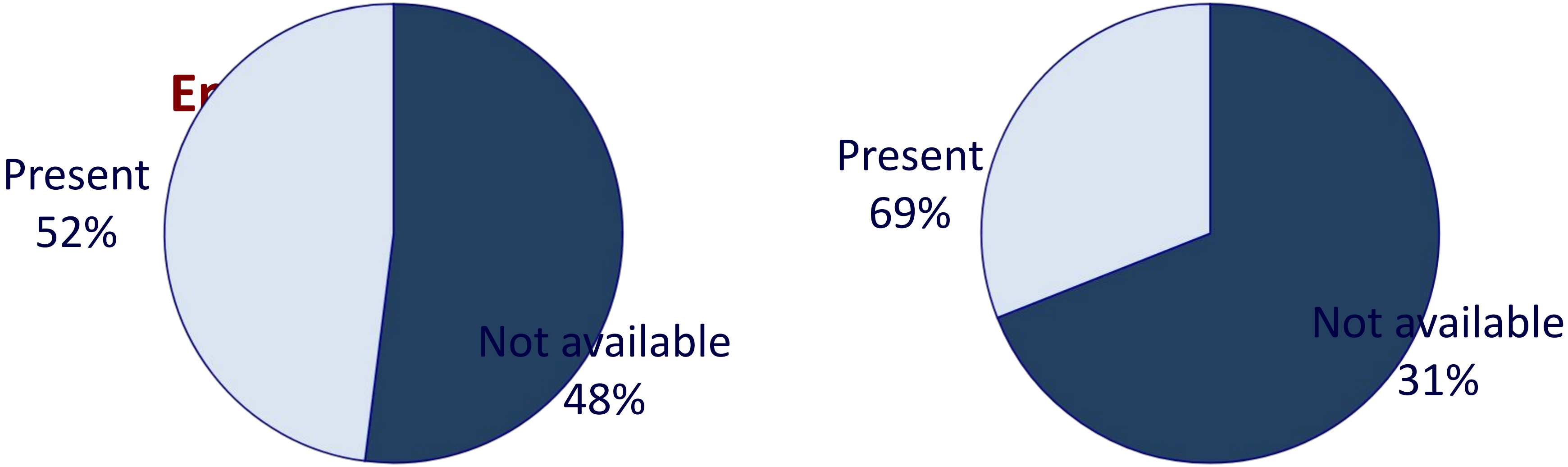
Specialists involved in the long-term care of a child with DSD





Multidisciplinary Teams in Specialist Centres

- An increasing number of DSD centres have access to specialist healthcare professionals
- However the majority of the centres do not have all staff available at the same time



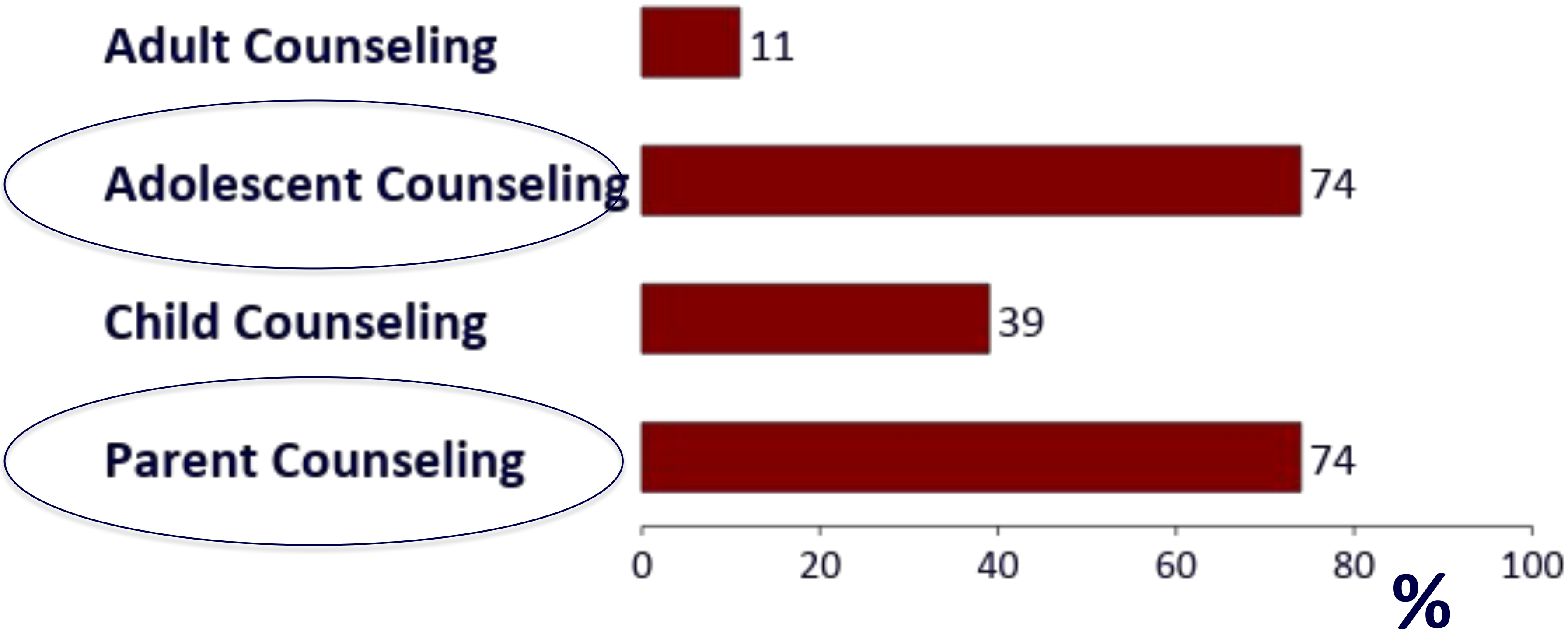
Specialists Not available

Current recommendation:

Psychosocial care should be an integral part of management

Psychology Support

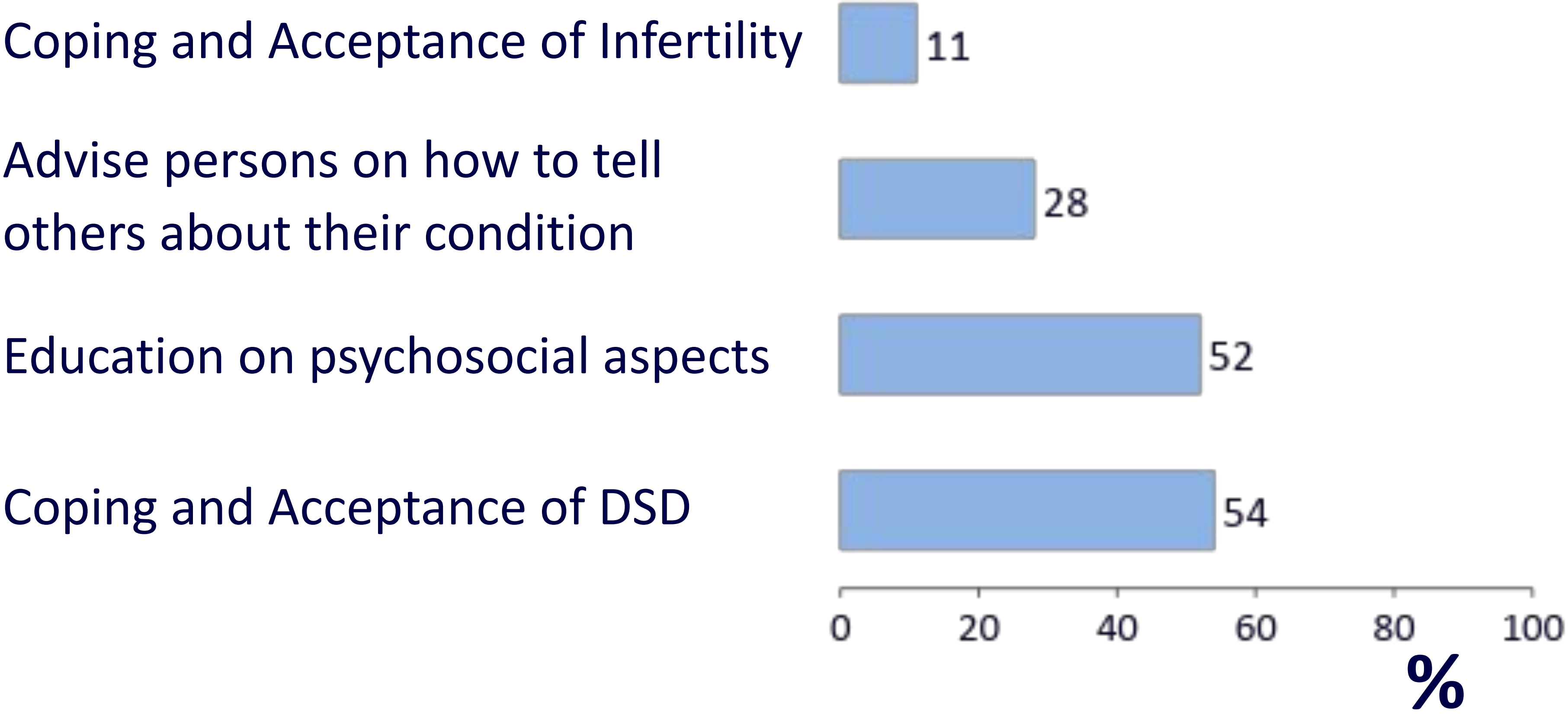
Involvement in Clinical care



Dessens AB et al., BMJ Paediatrics Open Aug 2017, 1 (1)

Psychology Support

Counseling activities



Dessens AB et al., BMJ Paediatrics Open Aug 2017, 1 (1)

Current surgical practice in DSD

In the past Surgery very early in life and to remove the gonads

Recent years There is a trend for delaying surgery or removal of gonads

Intersex activists have encouraged legislative bodies to ban elective irreversible “sex-correcting” surgery without the individuals’ consent

Current recommendation

Delay/Avoid unnecessary surgical procedures until the persons are mature enough to make their own decisions

Current surgical practice in DSD

A need to learn more about

- Risk for cancer in gonads if they are not removed
- Psychological development and well-being
- Hormones in adulthood
- Sexual functioning

Why is Research Important

Have you ever been in a situation where you had a question relevant to your condition, to which your doctor replied that there is no research available that allows him/her to answer your question?

- Cause
- Diagnosis
- Treatment options
- Outcome

There are many unsolved questions in our field

The aim of research is to learn more about DSD, find solutions to problems and ultimately improve clinical care and quality of life

Challenges in research in DSD

Requirements for conducting high quality research

1. The research topic should be important, with unsolved questions

Discrepancy between research topics proposed by doctors and by individuals with DSD – disagreement on what is more important

2. Sufficient number of participants in order to report reliable results

DSD includes a group of rare conditions: requires collaboration between centres at a national and international level

3. Individuals with DSD and healthcare professionals working together

Many individuals and families lost trust because they were mistreated in the past by healthcare professionals

Research priorities

- **Genetic causes of DSD**
- **Improvement in methods for diagnosis**

Research priorities

- **Studies related to long-term outcome – in adulthood**
  - Quality of life: sexual function, gender development, fertility
  - Long-term effects of treatment with hormones
  - Hormone treatment in adulthood (e.g. in menopause)
  - Other health problems: cardiovascular health, bone health
  - Cancer risk and development of reliable surveillance methods
- **Communication & Understanding between families and doctors**
- **Genetic causes of DSD**
- **Improvement in methods for diagnosis**

The information from the studies should be shared

with individuals and their families

Recent studies related to long-term outcome

Health status in 1040 adults with DSD: a European multicenter study.

Falhammar H et al. Endocrine Connections 2018 Mar;7(3):466-478

Hormone therapy and individuals’ satisfaction with treatment, in a large cohort of diverse DSD.

Nordenstrom A. et al. Clin Endocrinol (Oxf) 2018 Mar;88(3):397-408.

Recent studies related to long-term outcome

The Long-Term Outcome of boys with Partial Androgen Insensitivity Syndrome and a mutation in the Androgen receptor gene.

Lucas-Herald A, et al. J Clin Endocrinol Metab. 2016 Nov;101(11):3959-3967.

Bone mineral density in complete androgen insensitivity syndrome and the timing of gonadectomy.

King TJF, et al. Clin Endocrinol (Oxf) 2017 2017 Aug;87(2):136-140

It is necessary for individuals/families and professionals to work as research partners

Communication Of Doctors With Peer Support Groups And Families

A. Kyriakou[1], M. Rozas[2], A.B. Dessens[3], J. Bryce[1], V. Iotova[4], A. Juul[5], M. Krawczynski[6], A. Nordenskjöld[7], C. Sanders[8], O. Hiort[9], S.F. Ahmed[1]

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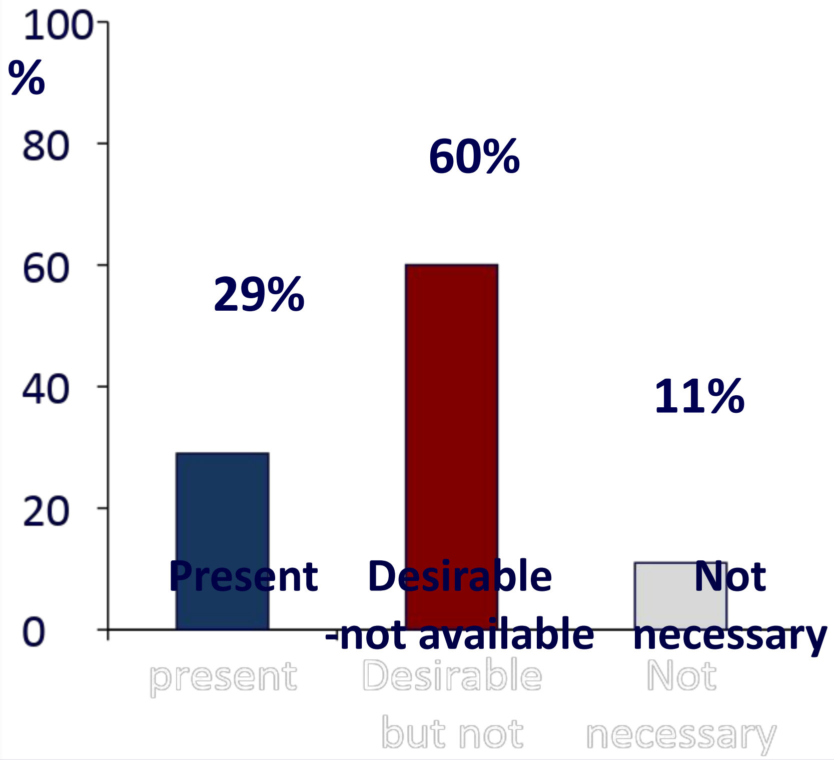
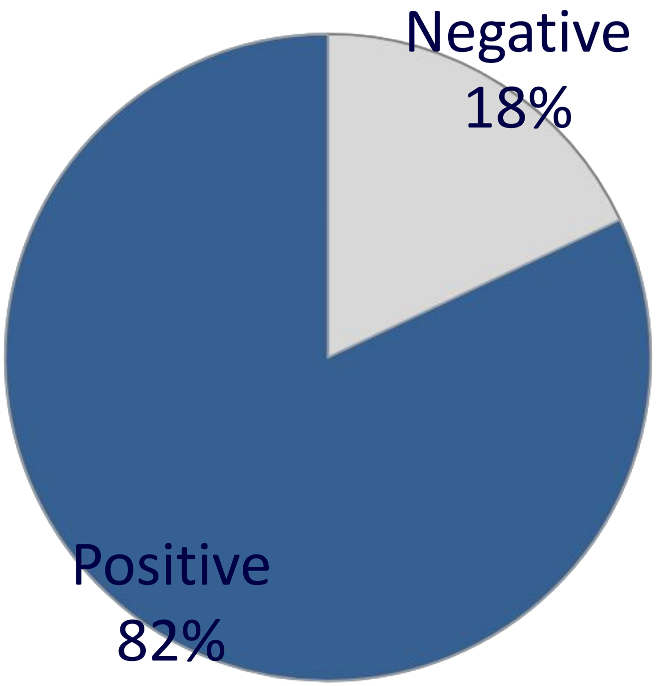
Communication of Doctors and Support Groups

Collaboration of healthcare professionals with Support Groups



Opinion for the Support Groups



Doctors should learn more about peer support groups for affected families in their area

International Survey of Support Groups for DSD

We have tried to ask 77 Support Groups worldwide about:

- The services they offer
- The information they provide
- Their opinion regarding the research topics that should be prioritised in the future
- If they collaborate with healthcare professionals
- If they collaborate with other Support Groups

International Survey of Support Groups for DSD

Only 16 Support Groups from 8 countries responded

The Netherlands DSDNederland

Klinefelter's Syndrome Association
Androgen Insensitivity Syndrome Support Group

United Kingdom

Germany

Eltern-Selbsthilfe XY-Frauen
Selbsthilfegruppe Eltern Intersexuelle Menschen

Sweden INIS

United States

Accord Alliance
Beautiful You MRKH Foundation, Inc
AIS DSD Support Group
Turner Syndrome Society
AXYS
AIS-DSD Support Group

Spain

GrApSIA

SIPD Uganda

Australia

Genetic Support Network of Victoria
Australian X & Y Spectrum Support

International Survey of Support Groups for DSD

Small number of responses – Not reliable results

How is the support provided?

- Mainly via social media, website
- All groups use phone calls, texts and emails
- A small number are using face to face discussions, either person to person or in group sessions

Information regarding the condition

- All groups provide information material (online or leaflets)
- They all agreed that they information material is not sufficient and it is not easily understood by the families

International Survey of Support Groups for DSD

- National meetings of families once a year, either by organising a social event or by organising a conference

- Informing society about DSD via social media and the press

- Collaboration with healthcare professionals

  - Support groups work very closely with Psychologists
  - They would wish for more regular contact with doctors

We also learnt

- Support Groups are run by volunteers
- There is no time available
- There is no money available

For working together, a questionnaire is not the right way to start

International Survey of Support Groups for DSD

- “Thank you for your contact. I am curious if you are aware that many advocates oppose the use of the term DSD to describe people with atypical sex characteristics”
- “Reducing intersex people to our body parts by calling us people with "Disorders" or "Differences" of sex development does not assist in us being viewed and treated as whole, equal human beings”

Comments from Support Groups

International Survey of Support Groups for DSD

“Unfortunately, after careful consideration, we have to decline to participate in this survey.

- The structure is too rigid to allow us to express our views, and is in fact tilted towards the interests of the medical establishment and not of the persons concerned.
- We are afraid this might be construed by some doctors as a justification to continue with research and treatments clearly in violation of ethics and human rights.
- May we suggest instead, that you could facilitate a meeting of support groups to facilitate a real discussion about the needs and interests of persons concerned and their organisations”

Comments from Support Groups

Workshop of persons/parents and professionals

Bologna, October 2016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Persons/Parents and a healthcare professional from 9 European countries (Austria, Belgium, Bulgaria, Germany, Ireland, Italy, Netherlands, Spain, United Kingdom)

Workshop of persons/parents and professionals

Organised by 2 Support Group members and a Nurse

Aims of the workshop:

- To hear the individuals’ and support groups’ voice
- To inform and educate professionals on the needs of individuals and families

Informal discussion, in groups

- Experience around the time of diagnosis
- Experience during childhood and adolescence
- Experience during transition to adulthood

? What is working well

? What is not working well

? Changes that need to be made in the future

Workshop of persons/parents and professionals

Experience around the time of diagnosis

- Worked well when professionals were able to help individuals and families *“find a language”* that was helpful to understand and to tell others

- *“Doctors liked my baby and told me that it is not a problem -families can live with it”*

- Does not work well when healthcare professionals lack knowledge about DSD
- Children or parents are not always informed about the diagnosis – kept as a secret.

Workshop of persons/parents and professionals

Experience during childhood and adolescence

Many individuals reported challenges to personal body integrity and a lack of respect

- especially linked to physical examinations from doctors

Workshop of persons/parents and professionals

Experience during transition to adulthood

- **Links to support groups, networks**;

  When persons found connections using social media and support groups their confidence was improved

- It is important for privacy to be respected and treated as a *"whole person"* especially when *"learning what my body **can do** rather than it cannot"*.

Moving forward

Lessons from Bologna

- **Organising Workshops locally**: by local professionals, in partnership with support groups/individuals with DSD /familes

- People want to be involved but need to be asked and invited
- Good environment = people will share

- Individuals/parents are not professionals and will not engage in the same way
- Relationships are the key, however it needs time to build connections

Moving forward

Working together

1. Networks between specialist centres for DSD
2. Partnership between Support Groups and healthcare professionals

European Reference Networks on Rare Endocrine Conditions (Endo-ERN)

Network of specialist centres for DSD

Sex Development and Maturation

Adult Endocrinologists

Paediatric Endocrinologists

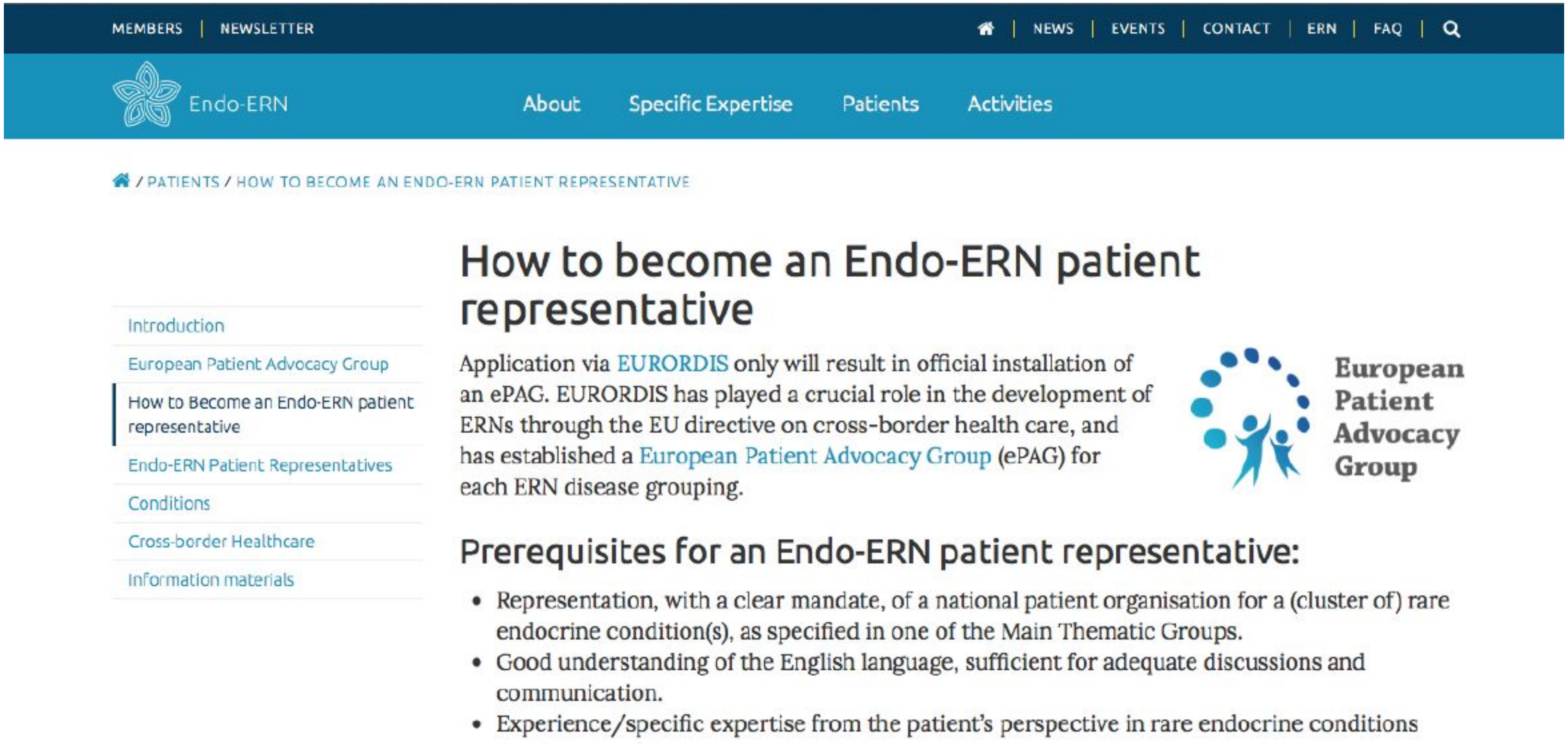
Elected representatives of individuals/families

European Patient Advocacy Groups (ePAGs)

https://endo-ern.eu

European
Patient
Advocacy
Group

Bring together elected representatives of support groups, to ensure that the individuals' voice is heard in the European Networks



How to become an Endo-ERN patient representative

Application via EURORDIS only will result in official installation of an ePAG. EURORDIS has played a crucial role in the development of ERNs through the EU directive on cross-border health care, and has established a European Patient Advocacy Group (ePAG) for each ERN disease grouping.

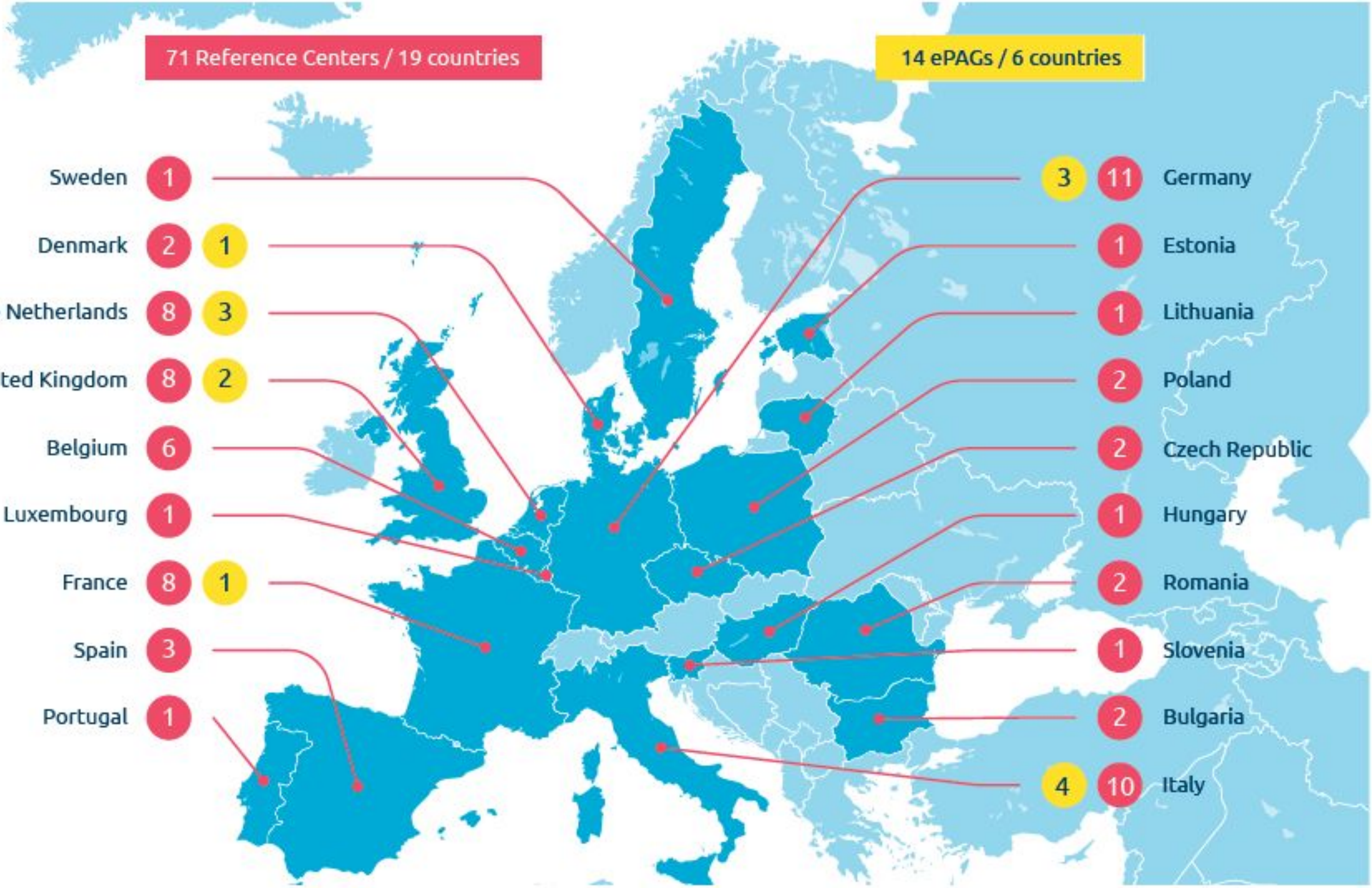
Prerequisites for an Endo-ERN patient representative:

- Representation, with a clear mandate, of a national patient organisation for a (cluster of) rare endocrine condition(s), as specified in one of the Main Thematic Groups.
- Good understanding of the English language, sufficient for adequate discussions and communication.
- Experience/specific expertise from the patient's perspective in rare endocrine conditions

Moving forward

Working together

Endo ERN– currently 71 centres



Grupo de
Apoyo a personas con
Síndrome de
Insensibilidad a los
Andrógenos y condiciones relacionadas

Yolanda

Marta

Carla Diaz Juhl

Thank you

Muchas Gracias