International Joint Recommendations
to address specific needs of
Undiagnosed Rare Disease Patients

CNA/CEF Annual meeting 3rd Nov 2016
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Overview

- Background
- Recommendations
- Dissemination
- Discussion on communication and future plans/activities

Background – Recognising the needs of the Undiagnosed population

- Needs of undiagnosed population at the heart of the rare disease community
- Highlight needs of the undiagnosed population and importance for EURORDIS to consider position and develop future advocacy actions
- Undiagnosed patients – cross-cutting all ERNs

Background – Undiagnosed Diseases Network international (UDNI)

- International network of clinical centres initiated by Helena and Mikk Cederroth (Wilhelm Foundation) together with William Gahl (NIH UDP)
- Modelled on the NIH Undiagnosed Disease Program (NIH **UDP**), USA (2008) and Network **UDN** (2013)
- Rome, 2014, Budapest, 2015, Vienna, Feb 2016 and Tokyo Nov 2016 conferences on rare diseases and undiagnosed conditions co-funded by Common Fund at NIH together with the Wilhelm Foundation

Objectives – Undiagnosed Diseases Network international (UDNI)

- Improve level of diagnosis and care for UD patients through development of common protocols
- Facilitate research into causes of UD by collecting and sharing data
- Create an integrated and collaborative research community across multiple clinical sites and among laboratory and clinical investigators

Structure – Undiagnosed Diseases Network international (UDNI)

- Consensus framework of principles, best practice and governance
- Board of Directors includes clinicians from Australia, Canada, Hungary, Italy, Japan and the US
- Fosters translation of research into medical practice
- International Patient advisory Group (iPAG), expected to play an important role in UDNI activities

UDNI meeting in Vienna, Feb 2016

- Consolidate the UDNI, discuss data sharing models within the consortium, membership criteria
  - 40 clinicians and scientists from USA, Australia, Japan, India and Europe
  - www.udninternational.org
- Patient organisations
  - EURORDIS, Wilhelm Foundation and NORD
  - Responsible for the development of the iPAG
    - Terms of Reference
    - Membership criteria
- Next meeting: 16-17 November 2016 in Tokyo

SWAN Europe first meeting 2016

- Envisioned objectives of a SWAN European network of specific patient organisations and support groups initiated by SWAN UK:
  - Networking and sharing approaches to supporting families of children/young people with undiagnosed genetic conditions.
  - Disseminating knowledge and facilitate access to genomic technologies within the undiagnosed community.
  - Building a platform for the undiagnosed community to have a voice in policy development in Europe.
  - Providing a forum to enable peer support between families of children/young people with undiagnosed genetic conditions.

SWAN Europe first meeting 2016

- Act as a point of contact for members of the research community to build connections with the undiagnosed community

- Organisations involved at the initiation stage:
  - SWAN UK, Hopen (Italy), ZON (Netherlands), *Sans Diagnostic et Unique (France) and Objetivo Diagnostico (Spain)*
  - Uniamo (Italy) and Filière AnDDi-Rares (french reference network for rare developmental abnormalities and intellectual disabilities)

Objectives of the recommendations

- Raise awareness of the needs and priorities of the undiagnosed disease patients
- Promote responsible and ethical international data sharing
- Advocate the needs to develop specific programmes at national and international levels
- Support the development and active engagement of an international network of patient representatives

International joint recommendations

Approach

- Coordinate with co-signatories their contributions to development of key messages
- Input from organisations and key individuals
  - ✓ “Interviewed” several national alliances, helplines and patient groups
  - ✓ Online communities - Creation of a dedicated RareConnect community
- Build on existing resources and ongoing initiatives:
  - ✓ SWAN UK/SWAN Europe
  - ✓ EuroGentest, ESHG, GA4GH
  - ✓ UK APPG Report (Rare Disease UK, GA UK, SWAN UK) “Undiagnosed” – Genetic conditions and the impact of genome sequencing

Recommendation (1)

Undiagnosed Rare Disease Patients should be recognised as a distinct population with specific unmet needs

- *Impact of diagnostic odyssey highlighted through several surveys (e.g. EURORDISCare, ERRADIAG, RareDisease UK: The Rare Reality)*
- *Not always a temporary situation – not delay healthcare provision*
- *Still very much an invisible and highly vulnerable population – priority to assess volume and needs in each country by national healthcare and social welfare authorities*

Recommendation (2)

National sustainable programmes dedicated for undiagnosed diseases should be developed and supported

- *Several existing multidisciplinary programmes and projects specifically set up to meet the needs of undiagnosed patients but disparity/inequality remain*
- *System to facilitate referral and access to next generation sequencing*
- *Sustainability*

Recommendation (3)

Knowledge and information sharing should be structured and coordinated (national and international)

- *Online community RareConnect platform – 8 languages*
- *Increase visibility of existing resources*
- *Significant role of umbrella organisations and helplines*
- *Developing partnerships between organisations and programmes – e.g. ASDU/AnDDi-Rares/MR Info service*

Recommendation (4)

Patients should be equally involved in governance of specific programmes and networks

- *UDNI (network of clinical centres) will include an international Patient Advisory Council*
- *Important role of PO in initiation, development and sustainability of specific programmes and networks*
- *Need to include patient representatives in governance of national undiagnosed disease programmes/projects*

Recommendation (5)

Ethical and responsible international data sharing should be promoted

- *Need widely accepted, should be implemented whilst respecting patients' preferences ("You should at least ask")*
- *Promising approach through matchmaking databases*
- *Encourage global participation in existing databases and projects such as the Matchmaker Exchange project*

Dissemination and Communication

- EURORDIS communication channels: social media; e-news and member news (started this week!)
- Presentation at the next conference of Undiagnosed Diseases Network International on 15th November in Tokyo
- Communication in November/December issue of Orphanews
- RareConnect undiagnosed community, SWAN Europe and dedicated patient organisations for undiagnosed
- Clinical leads of the different ERNs and ePAGS
- European Society of Human Genetics
- EuroGentest and other relevant European projects

We need your help!

- Promote the recommendations
- Development of protocols to inform and guide undiagnosed patients
- Share requests, concerns, “successes”
- Ensure effective communication
- Work with us to develop and coordinate activities specifically for undiagnosed rare disease patients

Thank you for you attention

- Paper now hosted in the EURORDIS library: http://www.eurordis.org/publication/international-joint-recommendations-address-specific-needs-undiagnosed-rare-disease-patients?platform=hootsuite

- Twitter: https://twitter.com/eurordis/status/793730848323428356 and https://twitter.com/eurordis/status/793731026841399296

- Facebook post: https://www.facebook.com/eurordis/posts/10154573709747707

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