



WP8: PATIENT ADVISORY COUNCIL: PATIENT ENGAGEMENT THROUGHOUT RD-CONNECT



Patient Advisory Council(PAC)

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- Voluntary working group coordinated by EURORDIS
- 16 active patient representatives
- 15 rare diseases represented
- 8 EU countries
- “Experts by experience” with wide range of experience and expertise on registries, biobanks, bioinformatics, data collection and sharing that help support project progress and highlight issues to be explored, discussed and considered.



Patient Advisory Council(PAC)

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Chris Sotirelis, UK Thalassaemia Society



Lydie Lemmonier, Vaincre la mucoviscidose (Cystic Fibrosis)



Joseph Irwin, Spinal Muscular Atrophy Support UK



Veronica Popa, Allan-Herndon-Dudley syndrome



Ciaran Scott, The Alkaptonuria Society



Alexandre Méjat, French Muscular Dystrophy Association - Téléthon



Virginie Bros-Facer, EURORDIS



Rainald von Gizycki, PRO RETINA Deutschland e.V.



Daniel Renault, Federation of European Associations of Patients affected by Renal Genetic Diseases



Dorthe Lykke, European Federation of Hereditary Spastic Paraplegia



Stefan Deckert, Deutsche-Heredo-Ataxie-Gesellschaft (DHAG)



Muriel Arcaute-Gevrey, CMT-France association (Charcot-Marie-Tooth disease)



Julian Isla, Dravet Syndrome Foundation



**Kay Parkinson
Director, Alstrom Syndrome Europe
CEO, Cambridge Rare Disease Network**



Marieke van Meel, NephcEurope



Sigurður Jóhannesson, Alternating Hemiplegia association of Iceland (AHCAI)/AHCFE Europe



Challenges??

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- PAC members are **volunteers** with busy lives
 - ▣ Always helpful to have flexible deadlines
 - ▣ Focus input on specific activities and interests

- Difficulty in ensuring a continuous and consistent engagement throughout the 6 years
 - ▣ Benefits in including new members progressively throughout such a long project
 - ▣ Diversity of background, experience and interests



Main achievements – ELSI with WP6

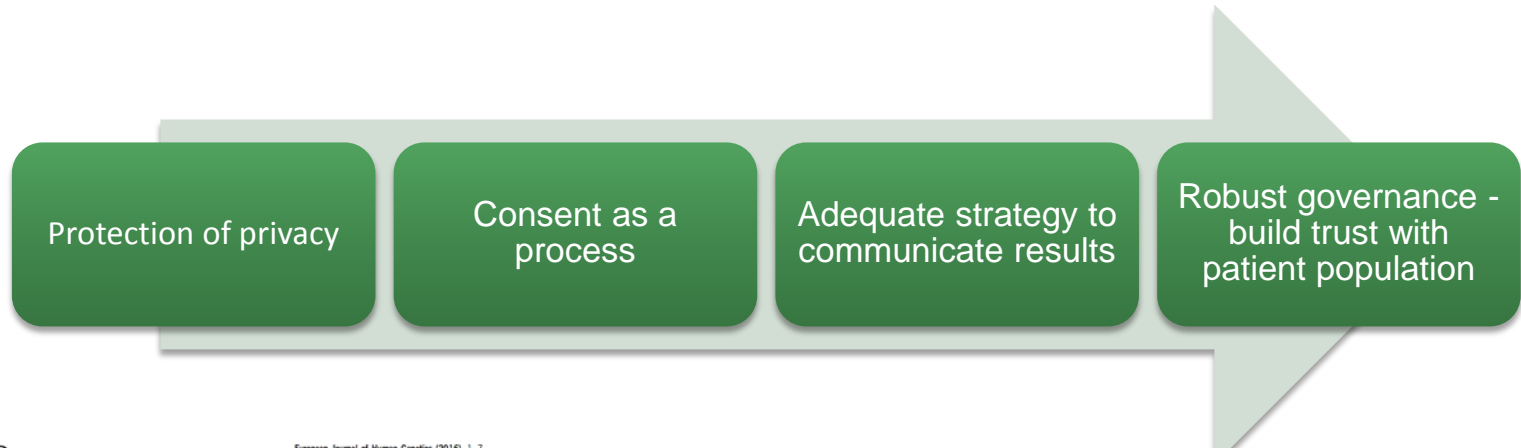
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- ❖ **Issues, concerns and perspectives surrounding data sharing and data privacy explored with RD patients:**
 - ✓ Focus groups (52 patients) – P. Mc Cormack
 - ✓ Delphi style exercise with ‘expert patients’ – 15 participants, 2 rounds. S. Courbier
 - ✓ Webinar on GDPR
 - ✓ Presentations at multiple workshops, meetings and conferences



Main achievements – ethical issues with WP6

❖ **Broad consensus about the necessity of sharing data internationally; benefits are always higher than the risks**



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ARTICLE

Improving the informed consent process in international collaborative rare disease research: effective consent for effective research

Sabina Gainotti^{1,9}, Cathy Turner², Simon Woods^{3,9}, Anna Kole^{4,9}, Pauline McCormack^{3,9}, Hanns Lochmüller^{2,9}, Olaf Riess⁵, Volker Straub⁵, Manuel Posada^{6,9}, Domenica Taruscio^{1,9} and Deborah Mascalonzi^{7,8,9}

European Journal of Human Genetics (2016), 1–6
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ARTICLE

‘You should at least ask’. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research

Pauline McCormack¹, Anna Kole², Sabina Gainotti³, Deborah Mascalonzi⁴, Caron Molster⁵, Hanns Lochmüller⁶ and Simon Woods¹



Main achievements- WP2 registries

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- **Integration in registry activities with WP2:**
 - Review on the “recommendations for improving quality of RD registries”
 - Poster – Kodra Y. et al (2017) RD-Connect annual meeting
 - Manuscript and upcoming publication
 - Participation in ISS Rare Disease Registry Summer School



Main achievements – WP3 biobanks

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- **Integration in Biobank activities with WP3:**
 - PAC member in Biobank Assessment Panel
 - EURORDIS is a member of BBMRI Stakeholders forum and liaise and feedback activities and discussions between the different projects and infrastructures (dissemination of RD-Connect activities)



For patients and families

The content of this section has been created by the rare disease patient representatives engaged in the RD-Connect work.



Patient representatives have a lot of knowledge about diseases and rare diseases. RD-connect uses this experience to help guide the research process.



Main achievements - communication

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- **Communication and dissemination with WP7:**
 - 5 full articles in RD-Connect Newsletter last year
 - Comprehensive glossary on terms used in the 3 projects and beyond
 - EURORDIS webinar RD-Connect (Summer School Alumni) with Libby and Sergi in February this year
 - Videos testimonials of PAC members on their involvement in RD-Connect (to be filmed later today...)



Plans for the next 6 months



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A EURORDIS INITIATIVE

- Right now: Data sharing/Data protection: EURORDIS Rare Barometer Voices - Large quantitative survey to gather perspective of RD patients (suggested by SAB)
- Plans for a short series of webinars on patient registries involving PAC/WP2 partners and external experts (ERNs)
- “Action list” of communication deliverables over the next 6 months for the website (who, what and when?)
- Develop a robust framework with PAC/PEC to sustain patient engagement in RD-Connect and its asset (registries, biobanks, platform)



Thank you!

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