

**WP8: PATIENT ENGAGEMENT  
THROUGHOUT RD-CONNECT  
VIA THE PATIENT ADVISORY  
COUNCIL**



# Patient Advisory Council(PAC)

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- Voluntary working group coordinated by EURORDIS
- 16 active patient representatives
- 15 diseases represented including Rare Kidney Diseases (EURenOmics), NMD (Neuromics) and others
- “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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**Joseph Irwin, Spinal Muscular Atrophy Support UK**



**Julian Isla, Dravet Syndrome Foundation**



**Virginie Bros-Facer, EURORDIS**



**Dorthe Lykke, European Federation of Hereditary Spastic Paraplegia**



**Jenny Versnel, Muscular Dystrophy UK**



**Elizabeth Vroom Duchenne Parent Project/United Parent Projects MD**



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



**Marieke van Meel, NephcEurope**



**Rainald von Gizycki, PRO RETINA Deutschland e.V.**



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



**Françoise Rouault, French Muscular Dystrophy Association - Téléthon**



**Lydie Lemmonier, Vaincre la mucoviscidose (Cystic Fibrosis)**



**Chris Sotirelis, UK Thalassaemia Society**



**Oliver Timmis, The Alkaptonuria Society**



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



**Veronica Popa, Allan-Herndon-Dudley syndrome**



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



# Main achievements – ethical issues with WP6

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ARTICLE

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

Sabina Gainotti<sup>\*,1,9</sup>, Cathy Turner<sup>2</sup>, Simon Woods<sup>3,9</sup>, Anna Kole<sup>4,9</sup>, Pauline McCormack<sup>3,9</sup>, Hanns Lochmüller<sup>2,9</sup>, Olaf Riess<sup>5</sup>, Volker Straub<sup>2</sup>, Manuel Posada<sup>6,9</sup>, Domenica Taruscio<sup>1,9</sup> and Deborah Mascalzoni<sup>7,8,9</sup>

European Journal of Human Genetics (2016), 1–6  
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[www.nature.com/ejhg](http://www.nature.com/ejhg)

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 McCormack<sup>\*,1</sup>, Anna Kole<sup>2</sup>, Sabina Gainotti<sup>3</sup>, Deborah Mascalzoni<sup>4</sup>, Caron Molster<sup>5</sup>, Hanns Lochmüller<sup>6</sup> and Simon Woods<sup>1</sup>



# Main achievements – ethical issues with WP6

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- ❑ **Data sharing and data privacy**
  - ✓ Methodology: Delphi style exercise with ‘expert patients’ – 15 participants, 2 rounds
- ❑ **Broad consensus about the necessity of sharing data internationally; benefits are always higher than the risks**
  - **Top priorities:**
    - ✓ Improve research for therapies
    - ✓ Develop collaborations between stakeholders
    - ✓ Improve diagnosis



# Main achievements – ethical issues with WP6

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- The main safeguard and incentive to encourage patients to participate in an international data sharing platform is having a **trustworthy stakeholder** as main curator of the data, including:
  - Government bodies
  - And/or Patient organisations
  
- The **role of patient organisations** is crucial:
  - ✓ In the management of the data sharing platform
  - ✓ To co-design the consent forms
  - ✓ To provide information to patients



# Main achievements- registries

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- **Integration in registry activities with WP2:**
  - Review and active input in quality self-assessment check list for registries
    - Poster – Kodra Y. et al (2017) RD-Connect annual meeting
  - PAC member driving integration of registry (inherited eye diseases) in ID-Cards
  - Presentation and dissemination at national and European workshops (registry workshop for patient groups, findacure UK; CHAFEA/Ciberer registry workshop)



# Main achievements - biobanks

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- **Integration in Biobank activities with WP3:**
  - PAC involved 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





# Main achievements - communication

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- **Communication and dissemination with WP7:**
  - Development of information materials for a new section on the RD-Connect website « for patients and families »
    - Video interview of PAC members on their involvement in RD-Connect
    - 5 full articles in the April edition of RD-Connect Newsletter
    - Comprehensive glossary on terms used in the 3 projects and beyond
    - Several ongoing activities to develop additional material including video interviews, tutorials, Q&As, animation on an overview of registries etc.



# Thank you for your attention

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