Patients needs & experiences

Simona Bellagambi
EURORDIS European Organisation Rare Diseases
www.eurordis.org
EURORDIS in brief

- Founded in 1997
- 734 member patient organisations
- 64 countries (26 EU countries)
- 38 National Alliances of RD Patients Organisations
- 51 European Federations of specific rare diseases
- Outreach to over 1800 patient groups
- Over 320 Volunteers: +75 patient advocates and +250 moderators
- 4 Million € Budget

An international non-profit, non-governmental umbrella rare disease patient organisation representing an estimated 30 million individuals in Europe whose mission is to be the voice of patients at European level.
RD Patient registries: an advocacy priority since 2006

- EURORDIS-NORD-CORD Joint Declaration of 10 key principles for RD patients registries (2012)
- Contribution to the EUCERD Core Recommendations on RD Patient Registration and Data Collection (2013)
- Contribution to the CEG-RD Recommendation on Ways to Improve Codification for RD in Health Information Systems (2014)
- EPIRARE Patient Survey (2012/2013)
- Partner in RD Connect (2012-2018)
EURORDIS-NORD-CORD Joint Declaration of 10
Key Principles for Rare Disease Patient Registries

- **Key instruments for advancing knowledge, research, care and treatments for RD**
- **First consensus into an international declaration by RD patient groups**
- **Underscore the importance of patient involvement in successful establishment, management and long-term maintenance of RDPR (patient needs, increase awareness, quality and quantity of data)**
- **Evidence that patient groups are very active and capable in this role complementing clinicians reported data (report on health related quality of life, satisfaction/utility of care and treatments – PM treatments outcomes, off label use outcomes)**
EURORDIS-NORD-CORD Joint Declaration of 10 Key Principles for Rare Disease Patient Registries

- Centred on a disease or group of diseases rather than a therapeutic intervention
- Interoperability (quality assurance and data security) and harmonization (exchange) between Rare Disease Patient Registries should be consistently pursued (CoEs, ERNs)
- A minimum set of Common Data Elements should be consistently used in all Rare Disease Patient Registries as those defined within the EUCERD JOINT ACTION WP8
- Rare Disease Patient Registries data should be linked with corresponding biobank data
EURORDIS/EPIRARE
Patient Survey on Registries (2012/2013)

• Specifically targeted at patients to gather their perspectives and expectations

• On line survey proposed in 11 languages, over 3,000 questionnaires analysed covering 500 diseases

• 14 questions including type of info, access modalities, governance and RD registers at national and European levels.
Diseases most represented

- Williams syndrome
- Behcet disease
- Scleroderma
- Duchenne muscular dystrophy
- Cystic fibrosis
- Rett syndrome
- Idiopathic achalasia
- Familial spastic paraplegia
- Idiopathic panuveitis
- Neurofibromatosis type 1
- Ehlers-Danlos syndrome type 1
- Proximal spinal muscular atrophy
- Tuberous sclerosis
- Beta-thalassemia
- Idiopathic steroid-sensitive nephrotic syndrome
- Systemic lupus erythematosus
- Prader-Willi syndrome
- Myasthenia gravis
- Epidermolysis bullosa
- Hereditary angioedema
EPIRARE Patient Survey: Aims of a register

Q: From the following list, please select the 3 aims that you think are the most important for a register.

- Description of the disease
- Epidemiological research (study of the frequency and distribution of the disease)
- Support for patient recruitment for clinical trials
- Surveillance of the patient population
- Evaluation and monitoring of the efficacy/safety of a treatment
- Genetic mutations database
- Healthcare and social services planning for patients
EPIRARE Patient Survey: Aims of a register

Q: From the following list, please select the 3 aims that you think are the most important for a register

- Description of the disease: 40%
- Epidemiological Research: 26%
- Support for patient recruitment: 24%
- Surveillance of the patient: 16%
- Evaluation/monitoring of the: 31%
- Genetic mutations database: 15%
- Healthcare and social service: 43%

02 September 2013 - Health Directorate at the Research DG of the European Commission
 Registers (professional) Survey VS Patient Survey:
Aims of a register

<table>
<thead>
<tr>
<th>Register reality</th>
<th>Patient expectations</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Epidemiological research</td>
<td>1. Healthcare/Social Services planning</td>
</tr>
<tr>
<td>2. Clinical research</td>
<td>2. Treatment evaluation (efficacy/safety)</td>
</tr>
<tr>
<td>3. Natural history of the disease</td>
<td>3. Natural history of the disease</td>
</tr>
<tr>
<td>4. Disease surveillance</td>
<td>4. Epidemiological research</td>
</tr>
<tr>
<td>5. Treatment evaluation (efficacy/safety)</td>
<td>5. Clinical research</td>
</tr>
</tbody>
</table>

02 September 2013 - Health Directorate at the Research DG of the European Commission
Q: From the following list, please select the 3 types of information you think are the most important to collect in a register

- Personal information (name, address, contact preferences, socio-demographic data…)
- Genetic information
- Medical information
- Medications, devices and health services used by the patient
- Patient reported outcomes (e.g. quality of life data, health status, etc.)
- Family medical history
- Patient participation to clinical research and bio-specimen donation
EPIRARE Patient Survey: Type of data

Q: From the following list, please select the 3 types of information you think are the most important to collect in a register

- Personal information
- Genetic information
- Medical information
- Medications, devices and health services used by the patient
- Patient reported outcomes
- Family medical history
- Patient participation to clinical research and bio-specimen donation

02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: Enrolment into a registry

Q: From the following list, please select the 3 most important types of information that should be communicated to the patient (relatives, guardians) before joining the register

- Register general aim and research objectives
- Type of users accessing the information contained in the register
- Right to withdraw
- Details on property rights regarding the information contained in the register
- Possibility of the register termination
- Possibility to be contacted or not for participating in clinical trials
- Name and contact of the register's manager
EPIRARE Patient Survey: Enrolment into a registry

Q: From the following list, please select the 3 most important types of information that should be communicated to the patient (relatives, guardians) before joining the register.

![Image of bar chart showing the percentages of selected information types.]

- Register general aim and research objectives: 65%
- Type of users accessing the information contained in the register: 33%
- Right to withdraw: 21%
- Details on property rights regarding the information contained in the register: 19%
- Possibility of the register termination: 4%
- Possibility to be contacted or not for participating in clinical trials: 34%
- Name and contact of the register’s manager: 14%

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EPIRARE Patient Survey: closure of the register

Q: How should previously collected information be handled if a register closes? It should be:

- Destroyed
- Stored indefinitely
- Stored for a limited time
- Made available to other registries or to the research community

02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: closure of the register

Q: How should previously collected information be handled if a register closes? It should be:

- Made available to other registries: 77%
- Stored indefinitely: 8%
- Stored for a limited time or to the research community: 8%
- Destroyed: 1%
EPIRARE Patient Survey: Access to data

• Q: In your opinion, who should have access to the information contained in the register?

  • Public Authorities
  • Public Institutions
  • Private institutions/citizens
  • Companies/Industries
  • Patients/Patients Associations
EPIRARE Patient Survey: Access to data

Q: In your opinion, who should have access to the information contained in the register?

Access to Register Data

- Public Authorities: 44%
- Public Institutions: 64%
- Private Institutions/Citizens: 27%
- Companies/Industries: 25%
- Patients/Patients: 89%
Q: If a participant wishes to withdraw from a register, what should happen to his/her data?

- Have his/her information anonymised for future research (irreversible destruction of participant’s identity)
- Withdraw the authorisation to future uses of his/her information
- Have his/her information destroyed
- Other
EPIRARE Patient Survey: Withdrawal from a registry

Q: If a participant wishes to withdraw from a register, what should happen to his/her data?

Withdrawal from a Register

- 68%: Have his/her information anonymised for future research
- 17%: Withdraw the authorisation to future uses of his/her information
- 23%: Have his/her information destroyed

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EPIRARE Patient Survey: Governance

Q: If a patient representative is a member of the register’s governing board, please indicate the importance of his/her opinion according to the domains of concern

- Financial and administrative issues
- Ethical and legal issues
- Information to be collected, research objectives
- Communication with the funding source, health care providers, patients, etc.
- Access to information contained in the register
- Coordination of all parties involved in the register
**EPIRARE Patient Survey: Governance**

Q: If a patient representative is a member of the register's governing board, please indicate the importance of his/her opinion according to the domains of concern.

<table>
<thead>
<tr>
<th>Domain</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Financial and administrative issues</td>
<td>51%</td>
</tr>
<tr>
<td>Ethical and legal issues</td>
<td>71%</td>
</tr>
<tr>
<td>Information to be collected, research objectives</td>
<td>76%</td>
</tr>
<tr>
<td>Communication with the funding source, health care providers, patients</td>
<td>64%</td>
</tr>
<tr>
<td>Access to information contained in the register</td>
<td>67%</td>
</tr>
<tr>
<td>Coordination of all parties involved in the register</td>
<td>60%</td>
</tr>
</tbody>
</table>

02 September 2013 - Health Directorate at the Research DG of the European Commission
Registers reality vs Patient expectations: Governance

EPIRARE Registries (professional) Survey - Q: Has the register a main governing board?

Only 29% have patient representatives

02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: Initiative for establishing the register

Q: If your disease has a register, please indicate by whom it was established

- Regional Authority
- National Authority
- University/Research Institute
- Hospital
- Patient Association
- Foundation
- Industry/Industrial Association
- EU Commission/EU Agency

02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: Initiative for establishing the register

Q: If your disease has a register, please indicate by whom it was established

02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: Financial sustainability

Q: Among the following funding sources, please indicate the 3 that could best assure the long term financial sustainability of the register

- Regional Authority
- National Authority
- University/Research Institute
- Hospital
- Patient Association
- Foundation
- Industry/Industrial Association
- EU Commission/EU Agency

02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: Financial sustainability

Q: Among the following funding sources, please indicate the 3 that could best assure the long term financial sustainability of the register.

![Diagram of Funding Sources for Long-term Financial Sustainability]

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Q: Do you agree that the European Commission should propose legislation to uniformly regulate rare disease patient registers across Europe?

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Uniform European Legislative Framework

- **84.8%** Yes
- **4.2%** No
- **11.0%** No opinion

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02 September 2013 - Health Directorate at the Research DG of the European Commission
EPIRARE Patient Survey: European Portal

Q: Do you find desirable that the European Commission and Member States build a common portal for all rare disease registers in Europe?

Common European Registry Infrastructure

- Yes: 90.7%
- No: 2.8%
- No opinion: 6.5%

02 September 2013 - Health Directorate at the Research DG of the European Commission
Conclusion 1/2

• Patients have a clear vision of the added value and benefits for them of a comprehensive European approach to Rare Disease Registers

• There is a gap between the purposes of registers in today’s reality and the expectations of patients

• Patients focus their interests in registers in view of disease knowledge production and care such as healthcare & social planning, treatment evaluation, natural history
Conclusion 2/2

• Patients unanimously demand that the legal aspects regarding Rare Disease Registers development be regulated at EU level

• Patients unanimously demand a EU platform primarily funded publicly (EU, National, Academic) involving patients in all aspects of Governance (scientific objectives, data to be collected, decision on the use of data, legal and ethical issues)

• Patients empowerment and capacity building – information, exchange of experience, networking, training - are needed for adequate and full involvement of patient representatives in the governance and activities of rare disease registers
European Platform on Rare Disease Registration: patients’ expectations 1/2

• Provide technical methods, tools, standards and support to encourage the creation of new registries and data collection networks and improving existing ones
  ➢ Specifically a **minimum common data set** (fundamental for the implementation of a global unique identifier), **quality standards**, **capacity building/training support** and **clear guidelines**

• Assure coherence with international RD initiatives involving registries activities
  ➢ Implementation of state-of-the-art sharing practices such as the use of **Global Unique Identifier** (RD Connect and now IRDiRC Task Force) to de-identify patient data while keeping the link to the corresponding data source.
  ➢ **Semantic operability**: choice of terminology such as those used by other related projects (e.g. HPO)
European Platform on Rare Disease Registration: patients’ expectations 2/2

• The success of the Platform will greatly depend on commitment of all stakeholders involved acting in a collaborative and flexible framework

• Serve as reference for technical, operational, ethical and social challenges by the concentration of efforts in this field and to address the uncertainty of the changing landscape
Considering an evolving registry ecosystem

• **European Reference Networks**: Registries will be at the centre of the ERNs so within healthcare infrastructure but will also need to feed into diagnostic and research infrastructures

• Research progress in disease areas lacking adequate natural history data, acceptable biomarkers, and valid endpoints will require mandatory interoperability between research registries and other research infrastructures such as biobanks and genetic databases (*RD Connect*)

• Targeted **data extraction from electronic health records** are likely to become a major source of data and the most cost-effective within the future of RD Registration
DATA FLOW

European Platform for Rare Disease Registries

- Centralised registry for very rare diseases
- Cross-talk with other European initiatives

De-identified Data Made Available

Global Rare Disease Community

- Healthcare professionals
- Patients and Carers
- Industry
- Researchers
- Patient Organisations
Expertise, health and data

A European Reference Network is more than the sum of its individual parts!

- Services will include delivery of specialist advice on diagnostic, care and treatment, for rare and complex cases.

- Specialist advice will be based on ‘collective experience, knowledge and expertise’ generated in the network, which is more than the sum of its individual healthcare providers.

Data sharing for healthcare and research:

- Data is our currency to exchange knowledge and learning, that drives improvements in outcomes and quality of life for our community.

- Data is the key to unlock the potential of these European Reference Networks.

- Safeguard our data. As caring for patients is caring for our data.

- When patients are separated from the ownership of their data, they data becomes vulnerable.
All patients expect data privacy …

Severity of disease

Level of concerns
Major Highlights of Patient Preferences

- Variability in preferences based on own experience, culture, age, disease severity and characteristics

- Patients see data sharing as an imperative as long as:
  - Consent is obtained respecting preferences
  - Protection of privacy and confidentiality is critical
  - Resulting progress is communicated
  - Trust established with data sharing network and patient representation in governance is demonstrated
  - Access to data transparent – especially cross boarder

- Can be achieved by:
  - Protection of privacy
  - Consent as a process
  - Adequate strategy to communicate results
  - Robust governance - build trust with patient population
Four aspects of data sharing

- Protection of privacy
- Securing consent
- Robust governance
- Feedback on use

Patient centered
Protection of privacy (1)

1. Protection of patient privacy and confidentiality of data is critical.
2. Patients and the community at large understand informational risks are involved in data sharing.
3. Risks should be mediated through safeguards (such as ethical review, informed consent and IT solutions) while maintaining/respecting reasonable time frames.
4. The patient community requires additional capacity building to be best informed during the consent process.

What the new rules say (GDPR – General Data Protection Regulation)

- Data Protection is a fundamental right of the individual.
- Health and genetic data are “sensitive” data deserving special protection.
- Safeguards include data minimisation, pseudonymisation and anonymisation when possible.
Securing consent (2)

1. Patients must consent to the sharing of their data
   - Consent models should provide research participants with the information they need to feel confident in participation
   - Consent models may vary depending on the type of data sharing and must be balanced with adequate communication with participants
   - Within a consent framework use of patient data is highly dependent on an ethical review. In the absence of a central European ethical review body a harmonised protocol should be strongly recommended in self-regulated transnational projects
   - The creation of consent models and materials should include real patient representation to encourage overall participation

What the new rules say (GDPR)

- Consent has to be given by a clear affirmative action establishing a freely given, specific, informed, specific, and unambiguous indication of agreement to data processing
- If a written declaration, request for consent must be distinguishable from other matters, in intelligible and accessible form, in clear and plain language
- Consent should cover all processing activities carried out for the same purpose
- Right to withdraw at any time
Robust governance (3)

1. A common regulatory framework can offer significant advantages
   - A common data sharing regulatory framework should facilitate data sharing and harmonise protection of sensitive data
   - The regulatory framework should include exceptions for health-related data that leave ample room for data sharing opportunities that propel scientific and medical progress

2. The Governance of data-sharing initiatives should include patients and be adaptive
   - Due to advances in regulations and technology, needs and expectations of all stakeholders are changing, including the patient community
   - The governance of data-sharing initiatives should reflect this need for adaptation
   - Patient representatives should be equally represented in the governance of data sharing networks

What the new rules say (GDPR)

→ GDPR harmonises rules across the EU but in a number of key areas Member States may keep or lay down specific national rules – for ex. on the protection of sensitive data. Harmonisation or fragmentation? What’s the impact on data sharing in ERNs?
→ Specific provision for use of data for scientific research. Anonymisation not compulsory
Feedback on use (4)

1. Outcomes of data-sharing activities should be communicated
2. Communication of outcomes to the patient community and the public at large should occur in a timely manner both at the aggregate and individual levels
3. Communication on individual results should be clearly anticipated in consent procedures

What the new rules say (GDPR)

→ Personal patient data can be reused for other purposes provided that his/her rights are ensured:
  - patients must be informed on other purposes,
  - must express NEW consent if previous for different purposes,
  - have the right to object to the new purpose
Patient Centred Approach (5)

1. Incentives for data-sharing should remain patient-centric
   - Data sharing culture change
   - Clear publication and data access policies (who get access, for what purpose, clear description of use and benefits)
   - Transparent collaboration with all stakeholders including industry
   - Should always keep patients' interest at their core
   - Communication of individual and aggregated results of the use of their data
   - Freedom of choice and personal preferences

What the new rules say (GDPR)

→ Change of culture also in GPRD with « data subject » holding a number of rights
   - Right of access to one’s own data
   - Right to be informed about these the purpose of the processing, the recipients of data;
   - Right to rectification of the data, to object the processing, and to restrict the processing of these data;
   - Right to receive a copy of the data and transport it (“data portability”);
   - Right to be forgotten

→ Whoever processes those data bears the burden of proof to show that rights have been respected
## Key messages

| Data is our currency to exchange knowledge and learning, that drives improvements in outcomes and quality of life for our community. |
|---|---|
| Data is the key to unlock the potential of these European Reference Networks. |
| Safeguard our data. As caring for patients is caring for our data. |
| The level of severity of disease directly relates to the level of concerns patients have to privacy. |
| Patients have clear rights on the use of their data that need to be integrated in the standard operating policies of ERNs. |

### Protection of patients privacy and confidential is critical AND is a fundamental right under the new regulation

- Protection of privacy
- Securing consent
- Robust governance
- Feedback on use

- Patients must give consent for the use and re-use of their data. Consent is an ongoing active process and not a single event.

- Communication on use and outcome of data is important and is mandatory to enable new purpose of use in ERNs.

Data sharing in an ERN should be patient centric in the governance and operational delivery of the networks. This will support increased harmonization across the network and aid data flow.
Thank you

Daniel - Sanfilippo syndrome