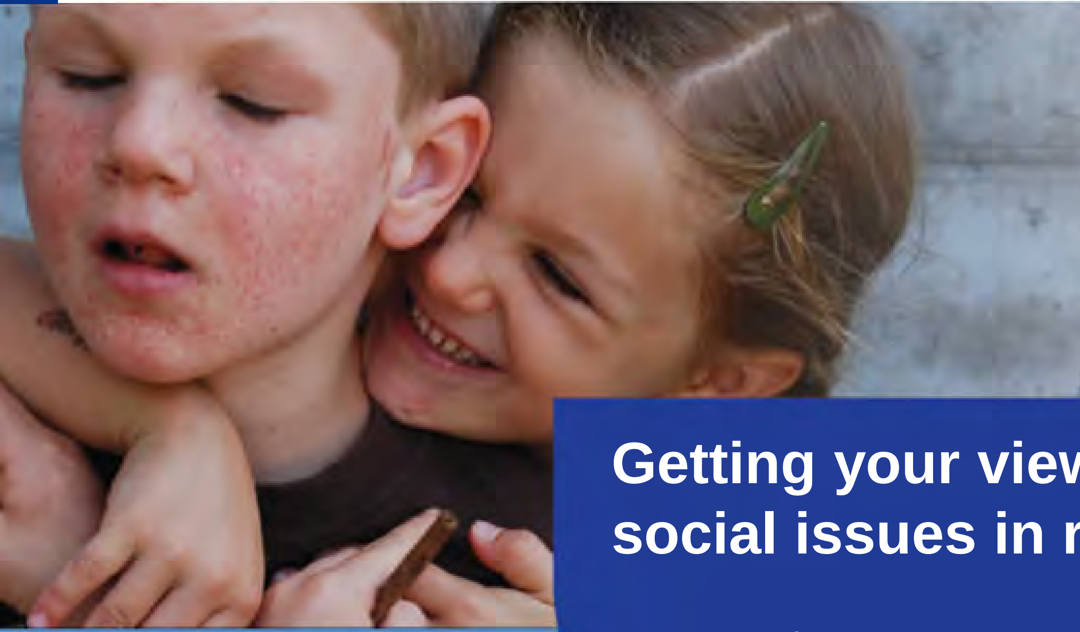
EURORDIS
Rare Diseases Europe



Getting your views on ethical, legal and social issues in research: Registries

Anna Kole, MPH – Registry and Biobanks Project Manager

What are patient registries?

- A patient registry is an **organized system** that uses observational study methods to **collect uniform data** (clinical and other) to evaluate specified outcomes for **a population** defined by a particular disease, condition, or exposure, and that serves a predetermined scientific, clinical, or policy purpose(s). The registry database is the file (or files) derived from the registry.

US Agency for Healthcare Research and Quality's (AHRQ) - Registries for Evaluating Patient Outcomes: A User's Guide

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Why are patient registries important for

- Patient registration addresses one of the key problems in the field of rare diseases - pulling information together from geographically and structurally dispersed sources, and making this information available for research and care purposes.

European Platform for Rare Disease Registries

- European Commission has announced strategic objective in creating a European Platform on Rare Diseases Registration at Joint Research Center (JRC) in Ispra, Italy
- Common services and tools for the existing (and future) rare disease registries in Europe.

European Commission

JOINT RESEARCH CENTRE

Institute for Health and Consumer Protection (IHCP)

EURORDIS Rare Diseases Europe

European Platform for Rare Disease Registries Project (EPIRARE)

- EURORDIS one of 11 project partners in the EPIRARE project aimed at building consensus and synergies for the EU registration of rare disease patients.

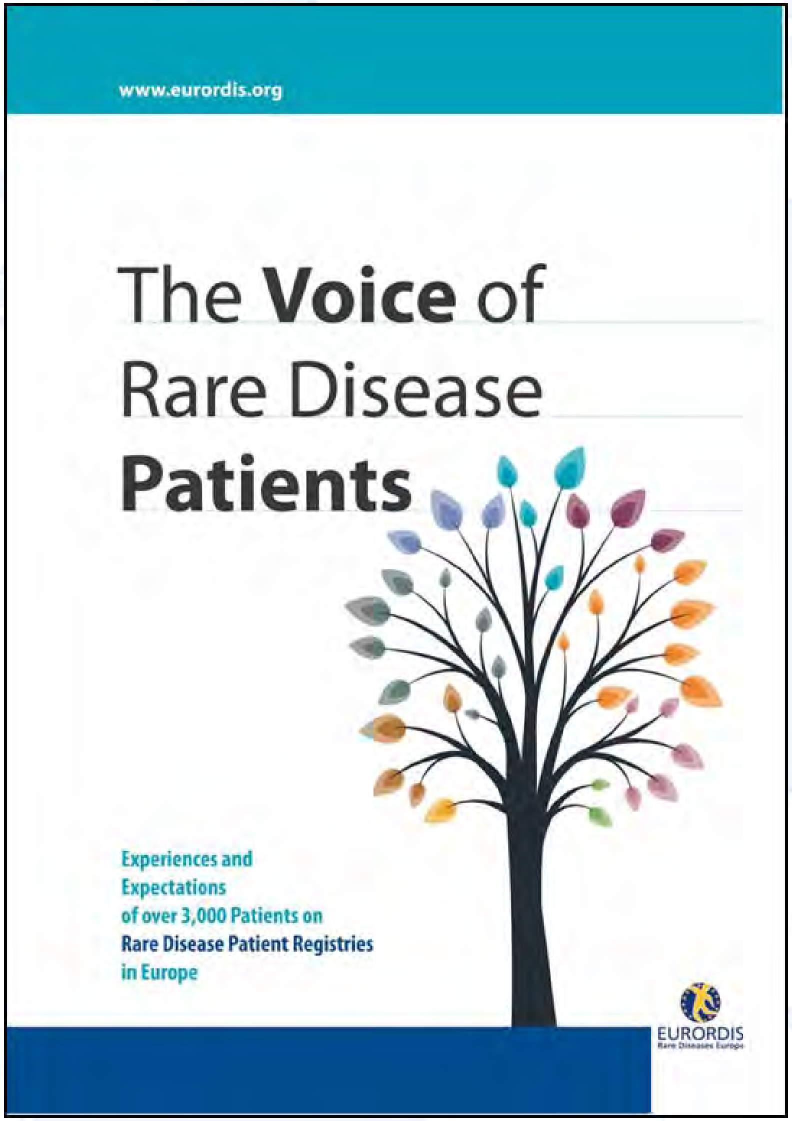
- Patient Survey to provide information on the experience and expectations of rare disease patient organisations and patients in registration and data

- Full list of deliverables available at http://www.epirare.eu/

Patient engagement

- Although rare disease patient registries are most often managed by universities, industry or public administrations, patient data ultimately belongs to patients.
- More and more patients now take an active role in initiating, designing, funding, and even directly collecting and sharing data within their own registry. Therefore, it is crucial and necessary to involve them actively in this process.

Patient Survey



www.eurordis.org

The **Voice** of
Rare Disease
Patients

Experiences and
Expectations
of over 3,000 Patients on
Rare Disease Patient Registries
in Europe

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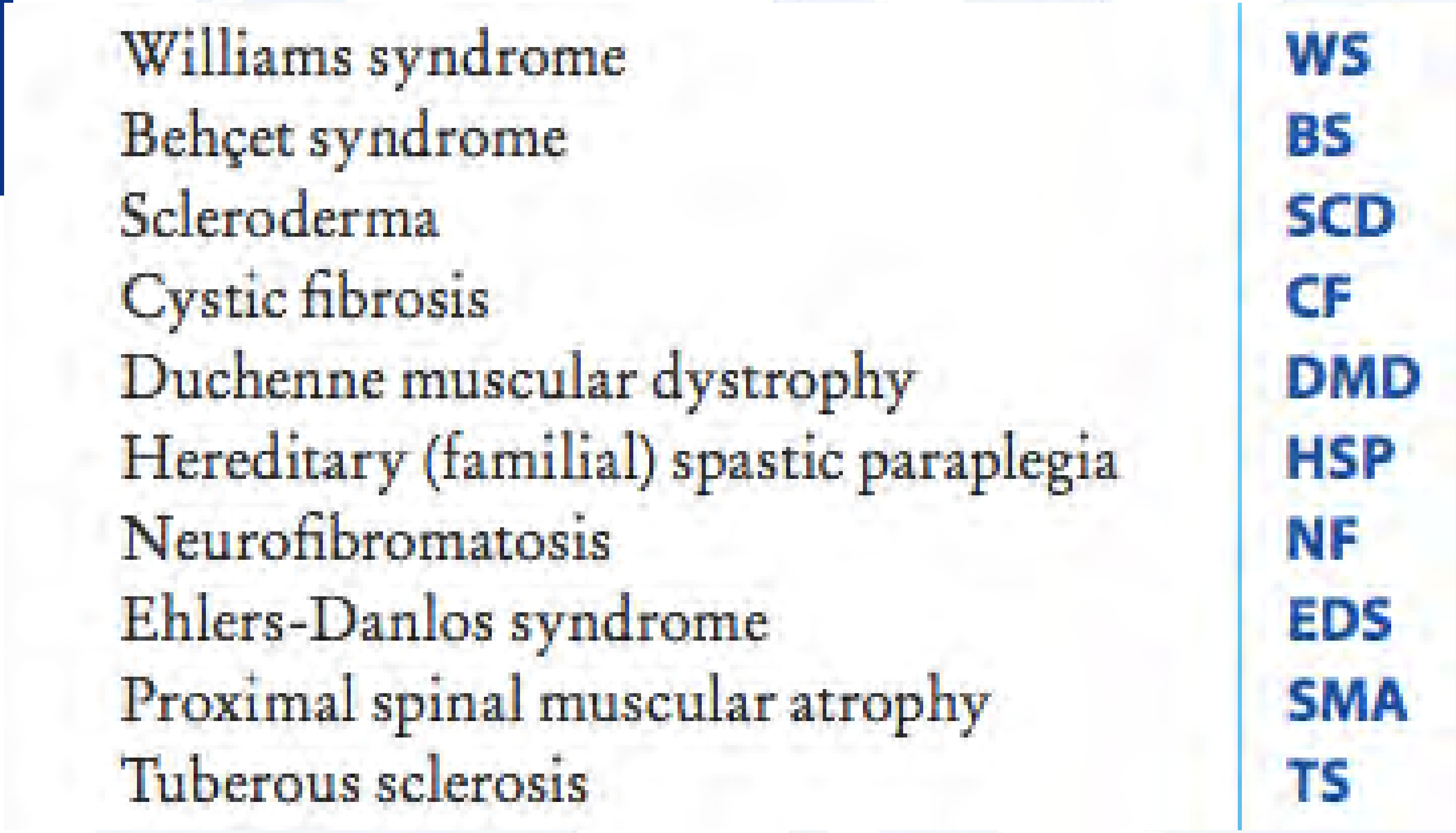
- July 1st, 2012 until February 1st,2013.
- Online, anonymous
- 10 languages

Respondents

- Overall 500 diseases represented (125 diseases represented 75% of responses)
- Overall 32 European countries represented (majority of responses from Spain, Italy, Germany, France, Greece, Portugal, Denmark, UK, Hungary, Czech Republic, Romania, Belgium)

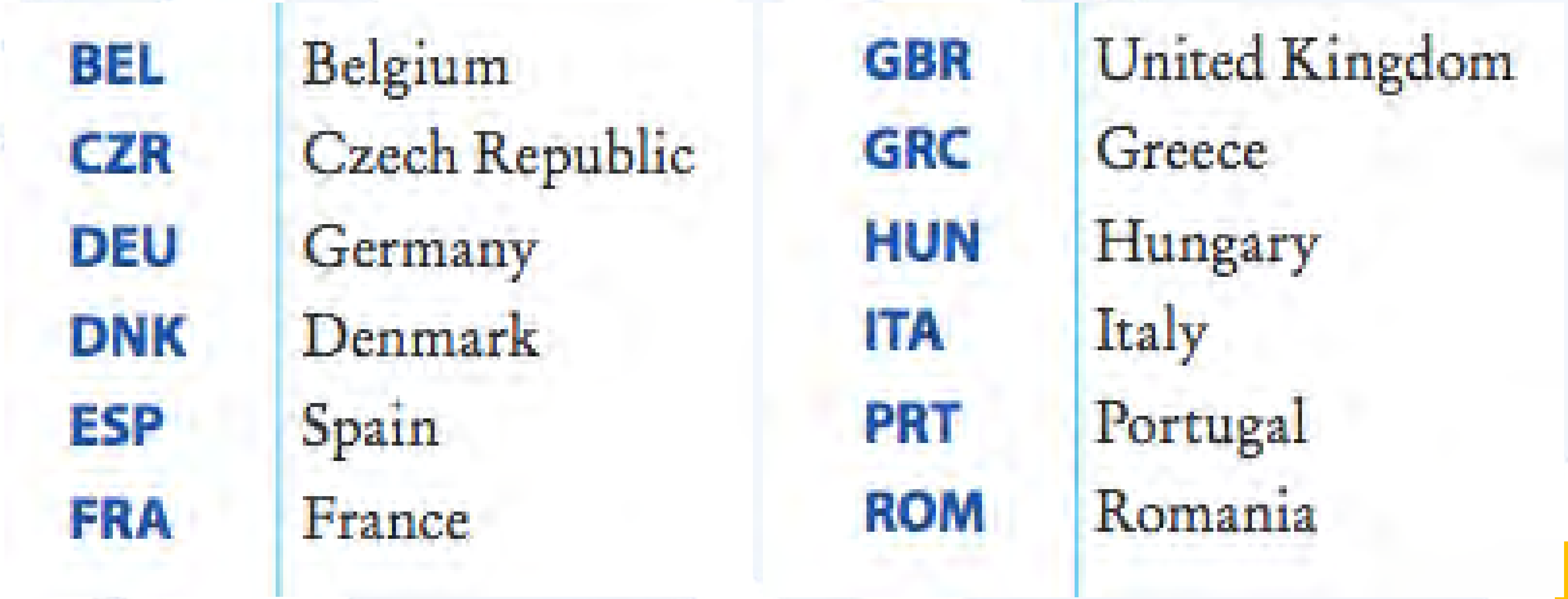
Results by Disease



Williams syndrome	WS
Behçet syndrome	BS
Scleroderma	SCD
Cystic fibrosis	CF
Duchenne muscular dystrophy	DMD
Hereditary (familial) spastic paraplegia	HSP
Neurofibromatosis	NF
Ehlers-Danlos syndrome	EDS
Proximal spinal muscular atrophy	SMA
Tuberous sclerosis	TS

> 50 responses

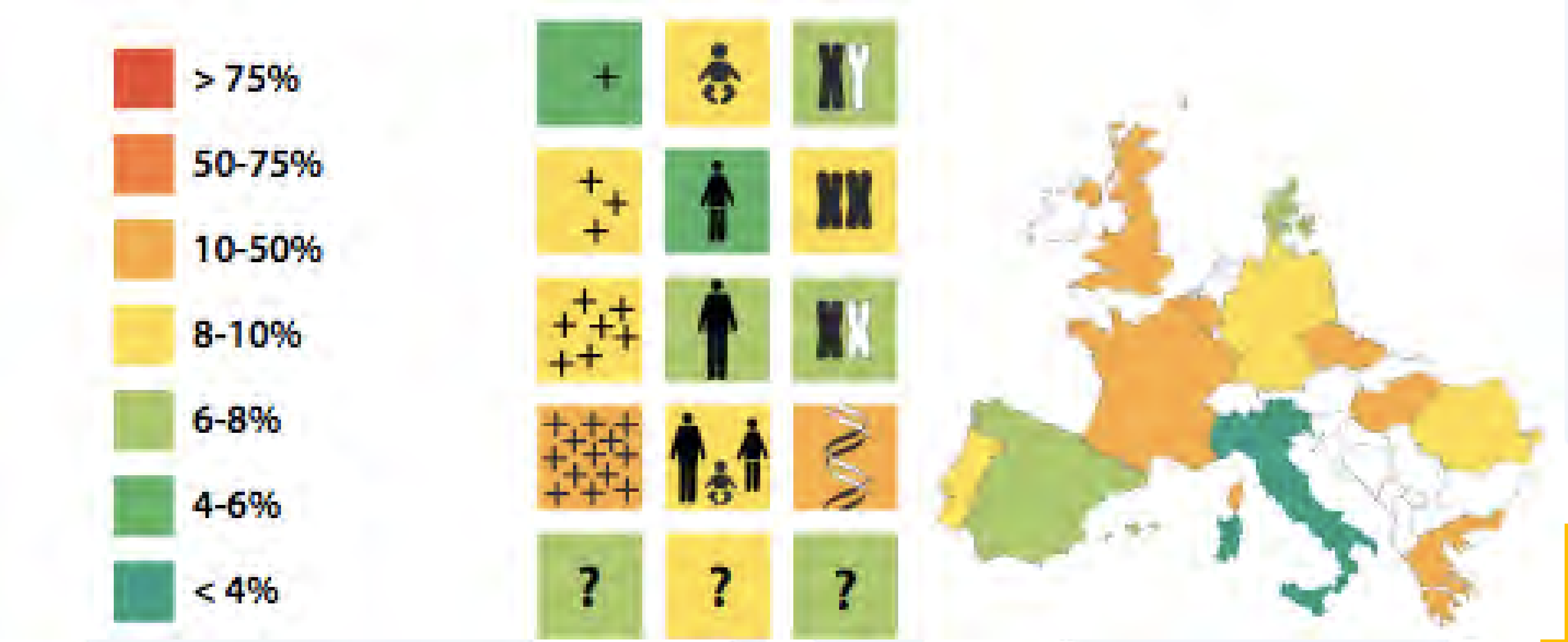
Results by Country



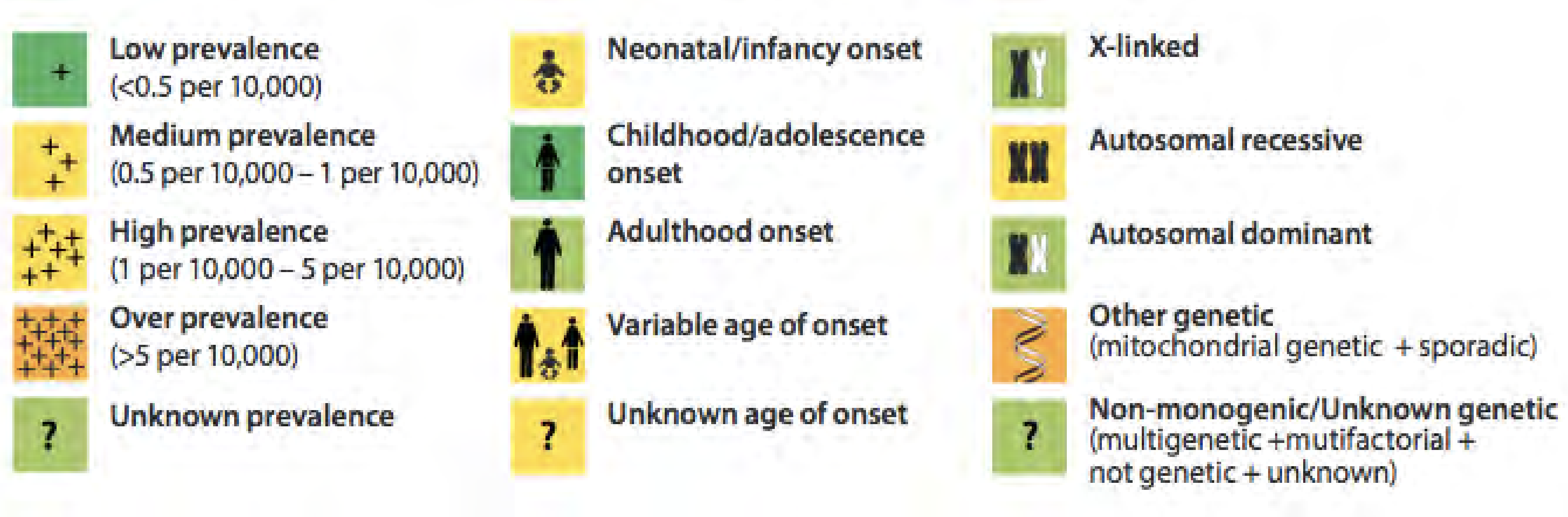
BEL	Belgium	**GBR**	United Kingdom
CZR	Czech Republic	**GRC**	Greece
DEU	Germany	**HUN**	Hungary
DNK	Denmark	**ITA**	Italy
ESP	Spain	**PRT**	Portugal
FRA	France	**ROM**	Romania

> 80 responses

Format of Overall Results

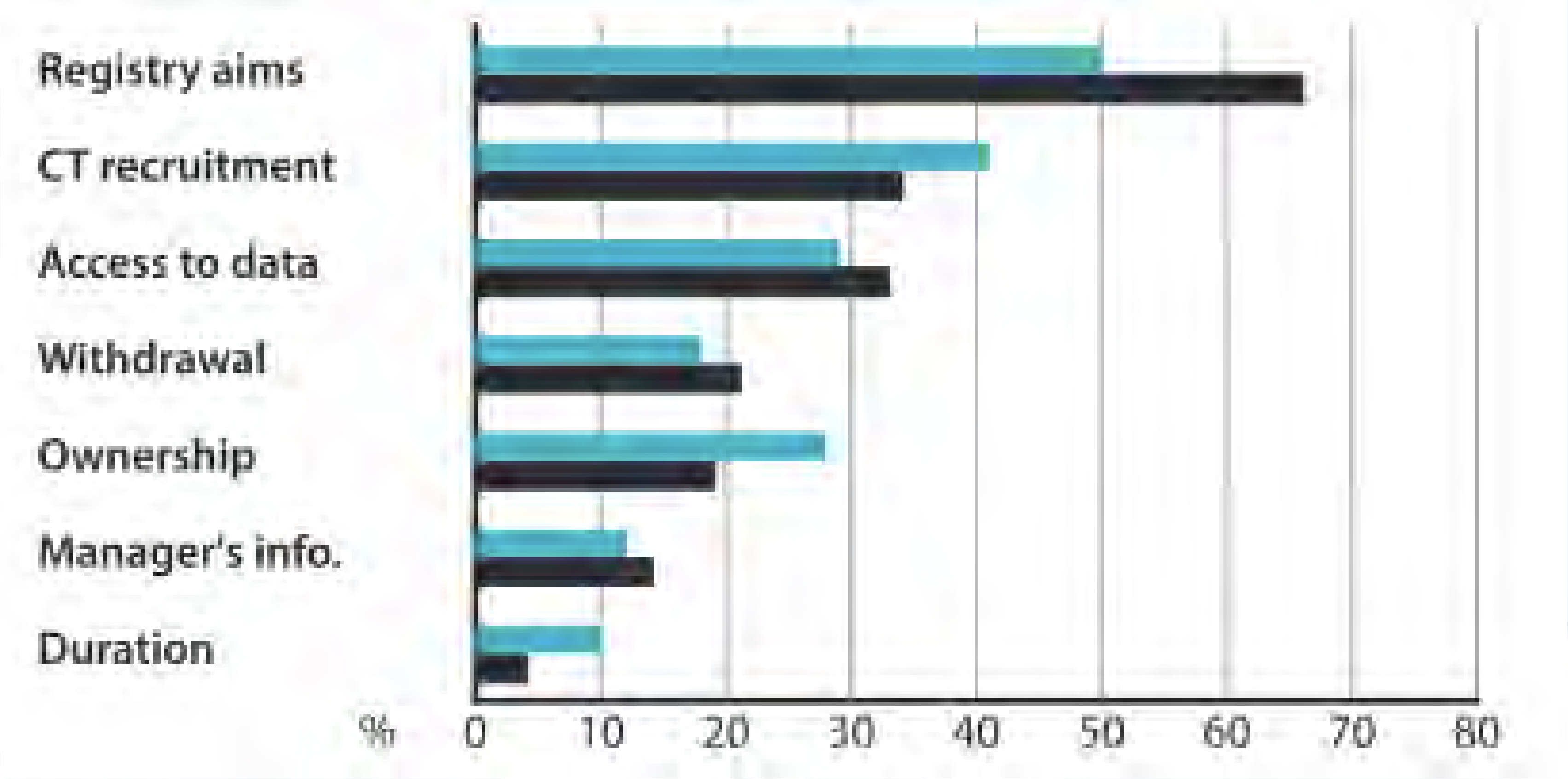


Results Legend



🟩 +	**Low prevalence** (<0.5 per 10,000)	🟨	**Neonatal/infancy onset**	🟩 XY	**X-linked**
🟨 + + +	**Medium prevalence** (0.5 per 10,000 – 1 per 10,000)	🟩	**Childhood/adolescence onset**	🟨 XX	**Autosomal recessive**
🟨 + + + + + + +	**High prevalence** (1 per 10,000 – 5 per 10,000)	🟩	**Adulthood onset**	🟩 XX	**Autosomal dominant**
🟧 + + + + + + + + + + + +	**Over prevalence** (>5 per 10,000)	🟨	**Variable age of onset**	🟧	**Other genetic** (mitochondrial genetic + sporadic)
🟩 ?	**Unknown prevalence**	🟨 ?	**Unknown age of onset**	🟩 ?	**Non-monogenic/Unknown genetic** (multigenetic +mutifactorial + not genetic + unknown)

Format of Specific Results



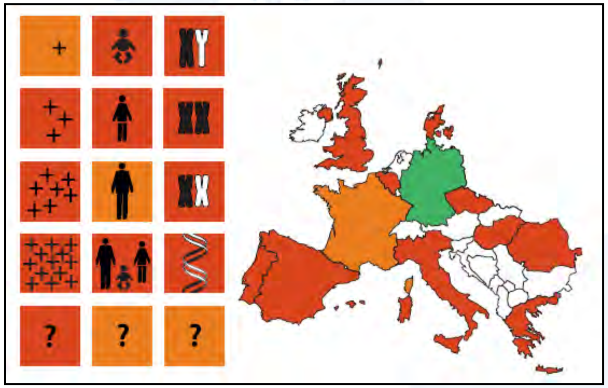
Overall Results - consensus

- Clear consensus on a number of issues illustrated by a high overall number of responses and little variability across country or disease groups.
- Questions regarding
  - Structural elements of a registry
  - Patient involvement in registries
  - Registry Governance and Sustainability

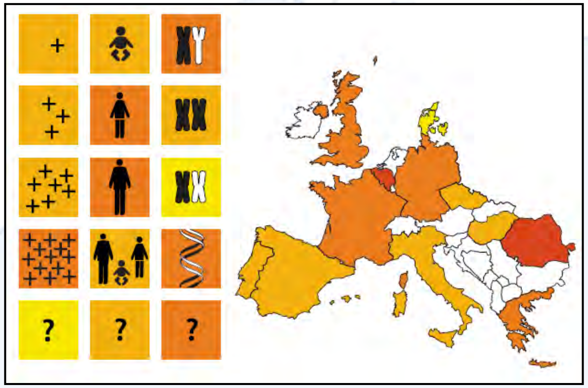
Structural Elements of a Registry

Registry Aims

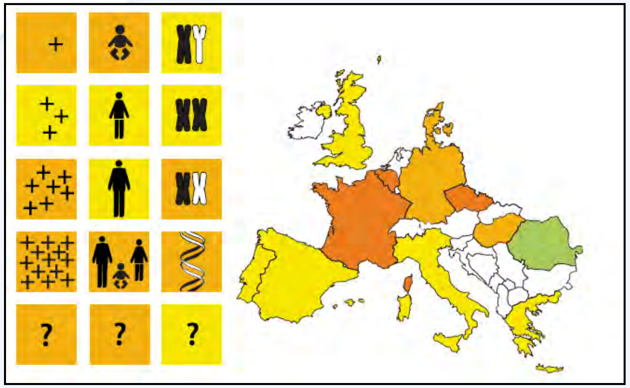
From the following list, please select the 3 aims that you think are the most important for a register. (Multiple choices, expressed as score)



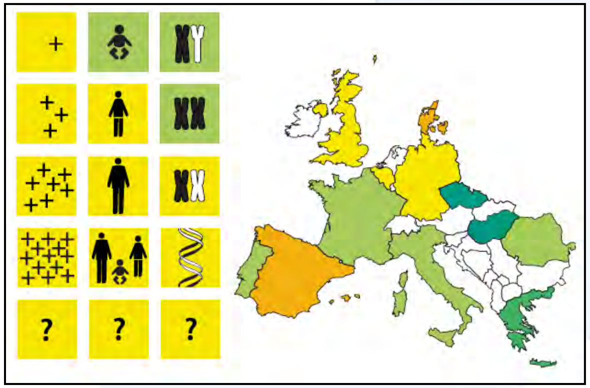
43% Healthcare and social planning



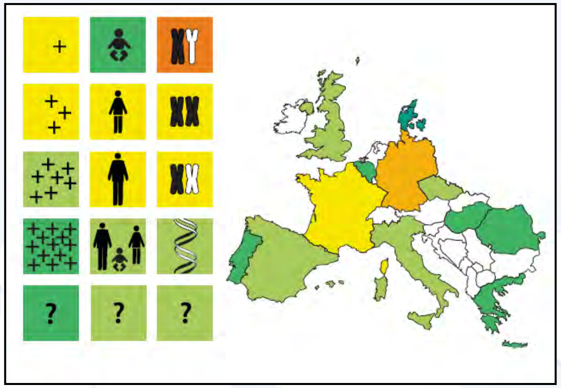
34% Evaluation and monitoring treatments



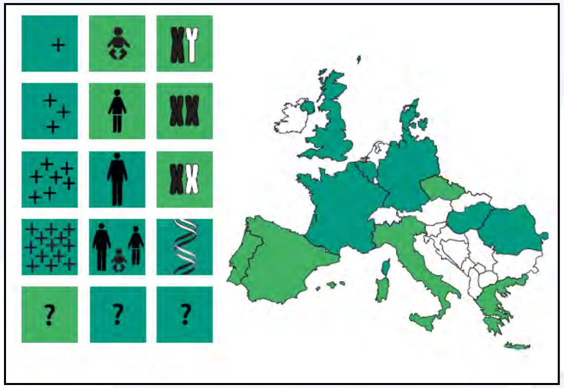
30% Description of the disease



26% Epidemiological research



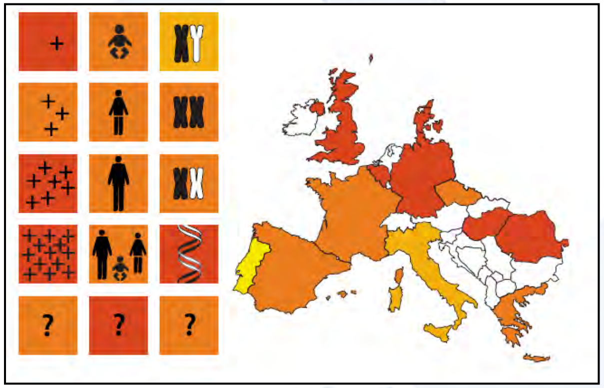
24% Recruitment clinical trials



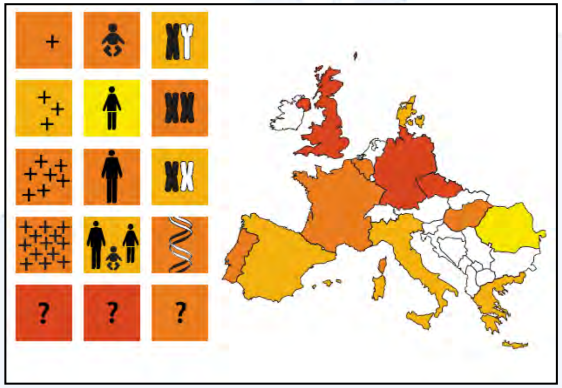
14% Genetic mutations

Type of data collected

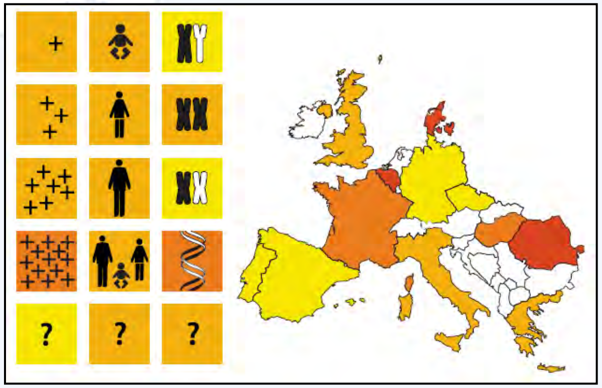
From the following list, please select the 3 types of information you think are the most important to collect in a register. (Multiple choices, expressed as score)



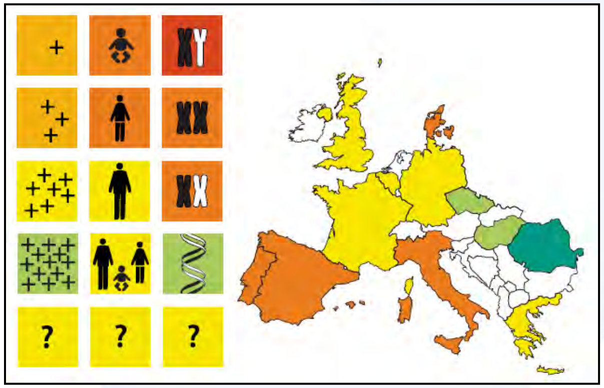
39% Medical Information



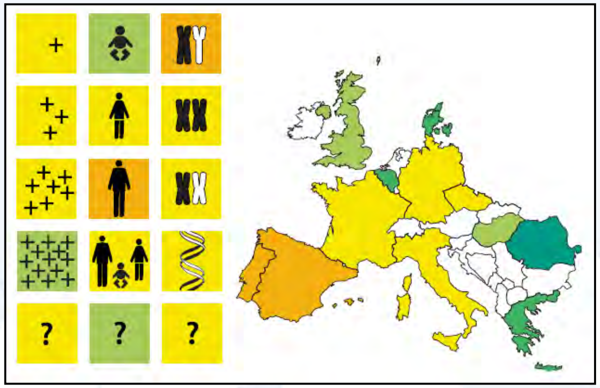
36% Patient-reported outcomes



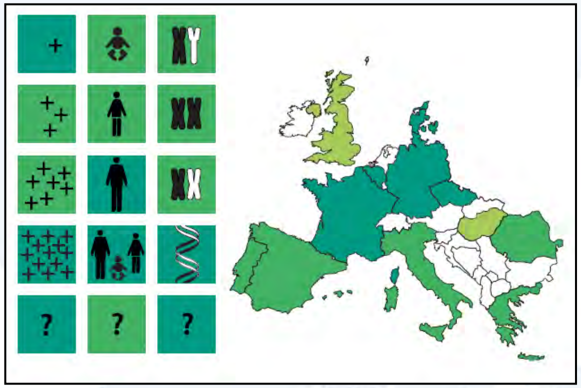
32% Therapeutic use



31% Genetic information



27% Participation in research and biobanks



15% Personal information

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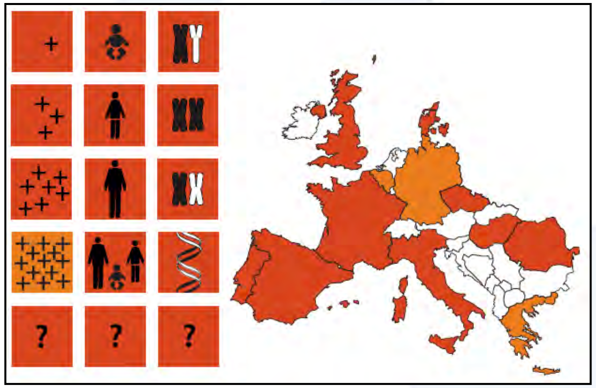
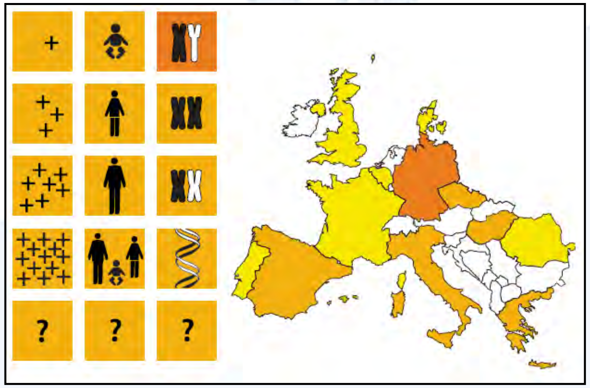
Patient Involvement in Registries

Info communicated upon enrollement

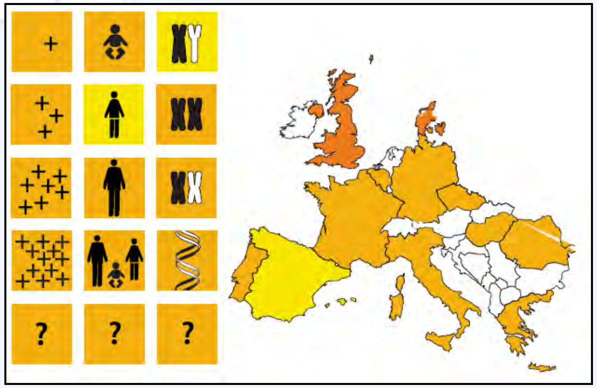
Please select the 3 most important types of information that should be communicated to the patient (relatives, guardians) before joining the register. (Multiple choices, expressed as score)



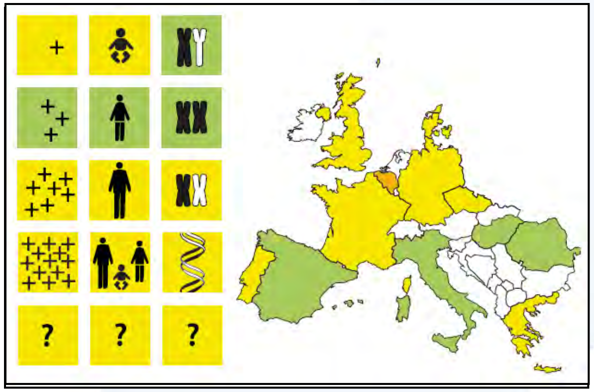
66% Registry aims



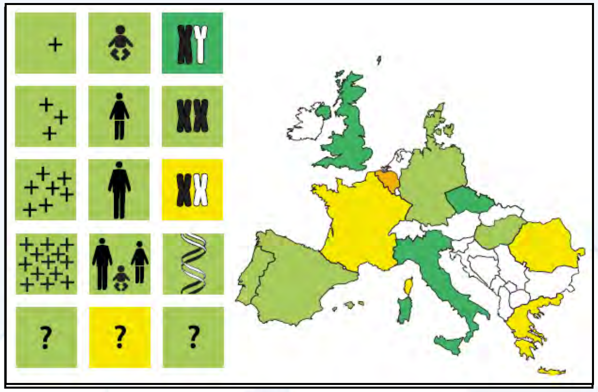
34% Recruitment for clinical trials



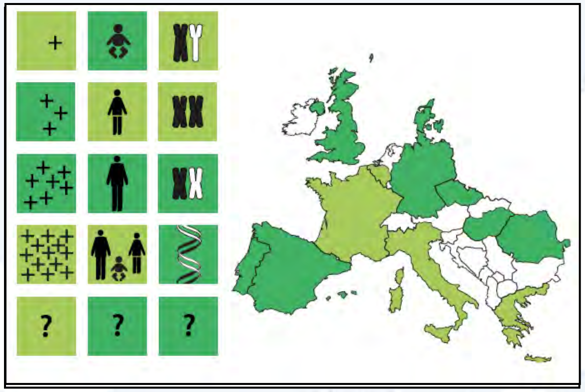
33% Access to data



21% Right to withdraw



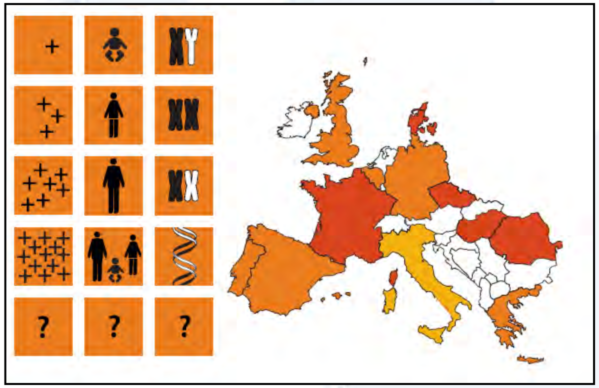
19% Data ownership



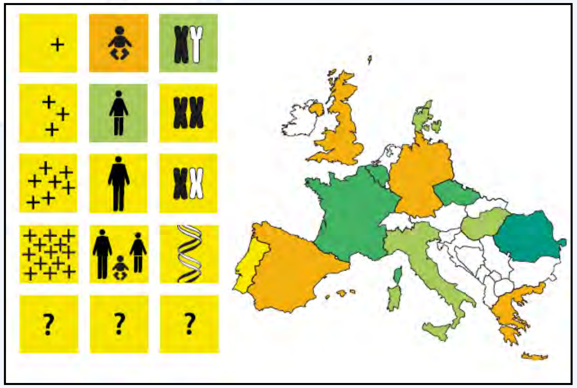
15% Registry contact info

Withdrawal from a registry

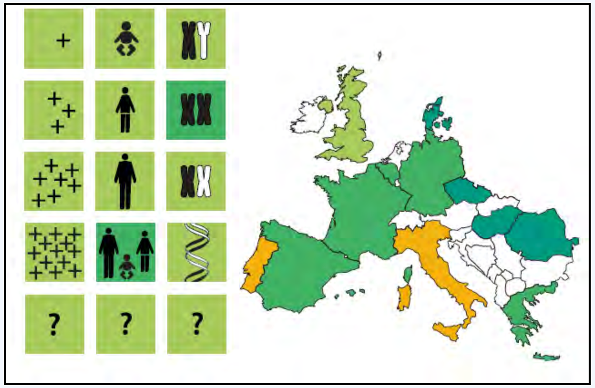
If a participant wishes to withdraw from a register, what should happen to his/her data? (Unique choice, expressed as percentage)



68% Data anonymised for future research



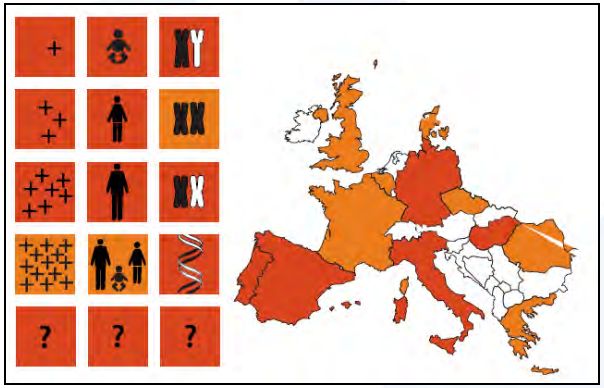
23% Data destroyed



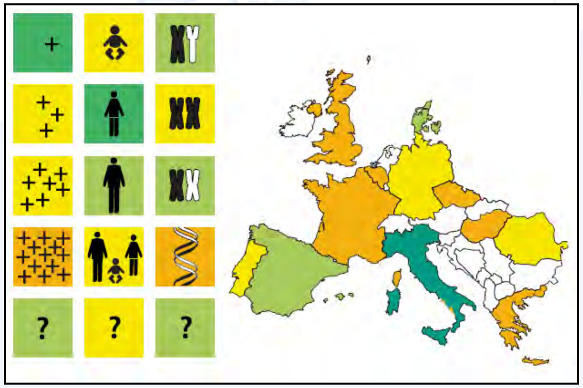
17% Authorisation withdrawn for future use

Registry closure

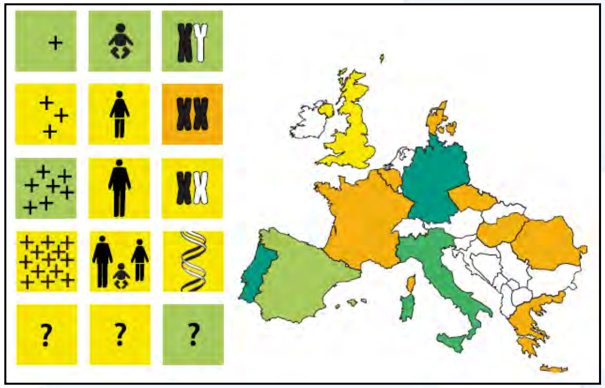
How should previously collected information be handled if a register closes? (Single choice, expressed as percentage)



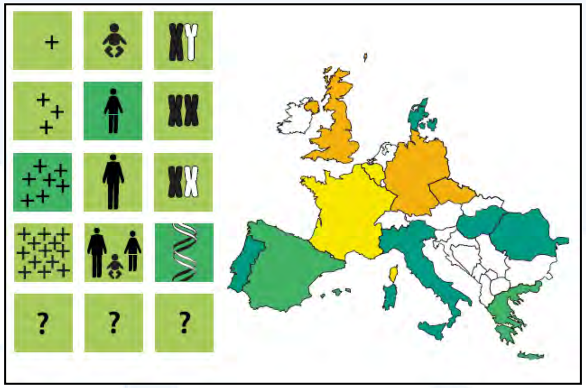
77% Data made available to other registry or research community



8% Data stored for a limited time



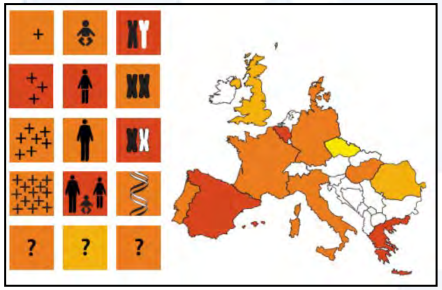
8% Data stored indefinitely



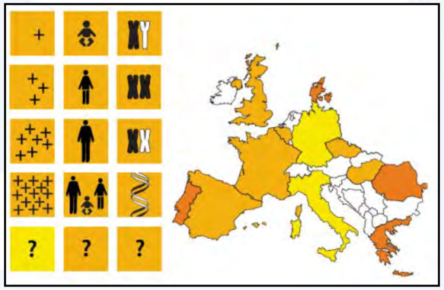
7% Data destroyed

Initiative for Establishing Registry

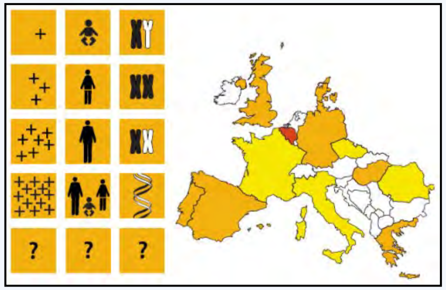
If your disease has a register, please indicate by whom it was established.
(Multiple choices, expressed as percentage)



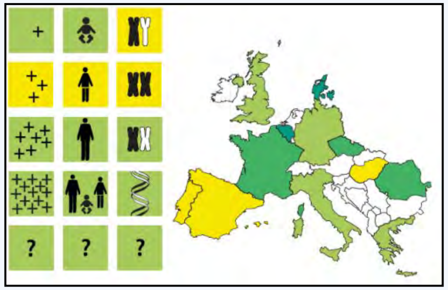
66% Patient organisation



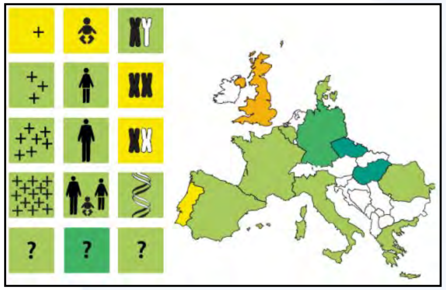
34% Hospital



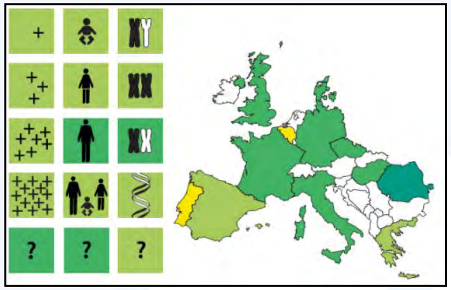
33% University/research institute



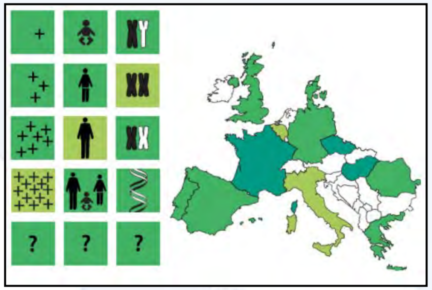
33% Foundation



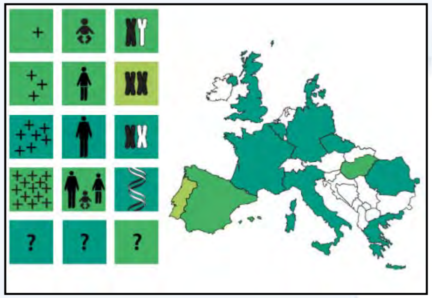
66% National authority



34% EU Commission/Agency



33% Regional authority

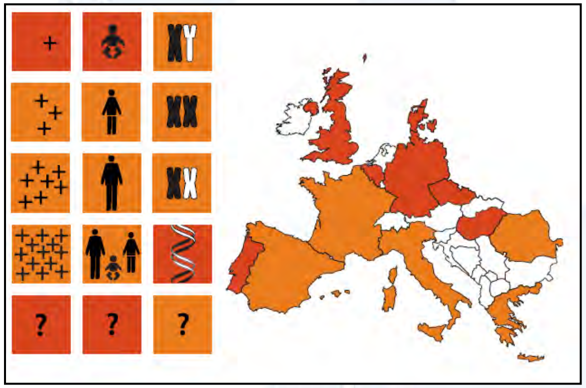


33% Industry

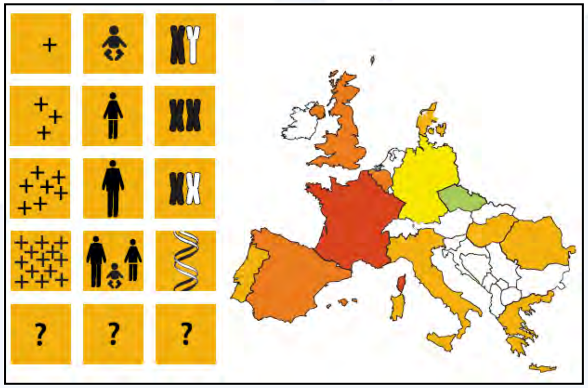
Registry Governance and Sustainability

Registry Users/Access

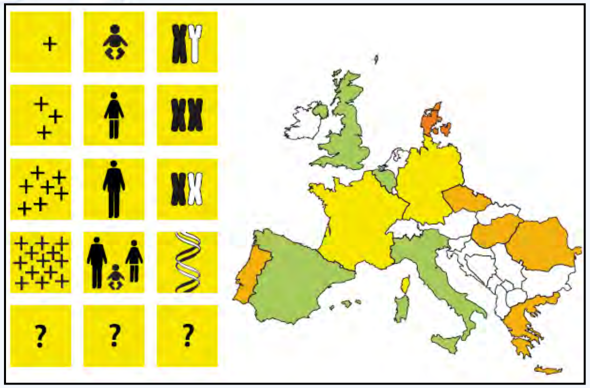
In your opinion, who should have access to the information contained in the register? (Multiple choices, expressed as percentage)



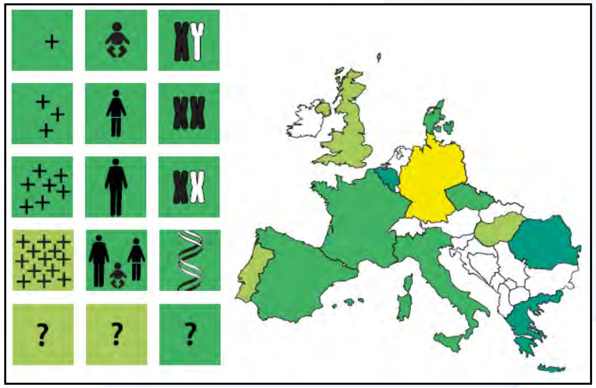
66% Patient organisations



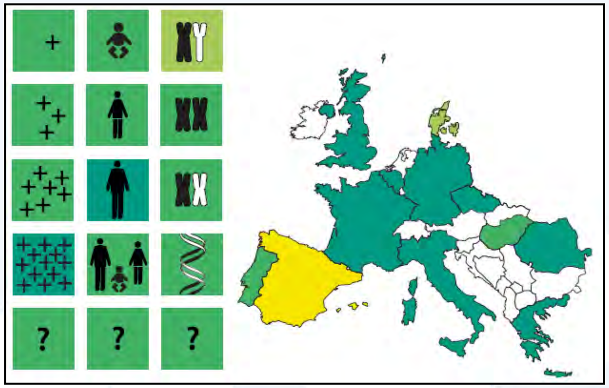
34% Public institutions



33% Public health authorities



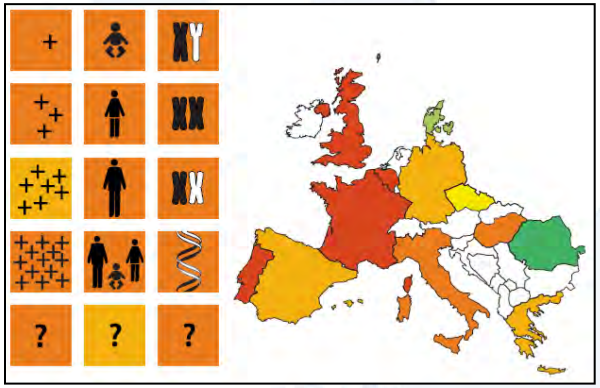
21% Private institutions/citizens



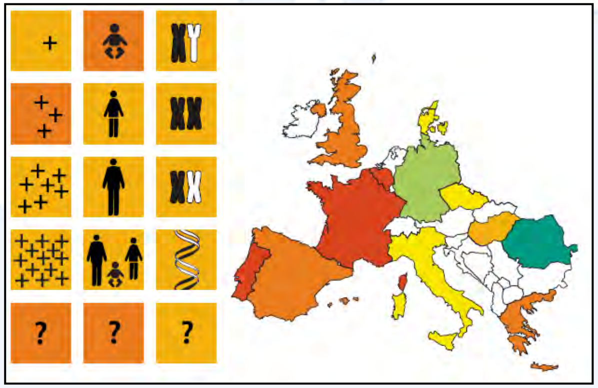
19% Industry

Registry Governance

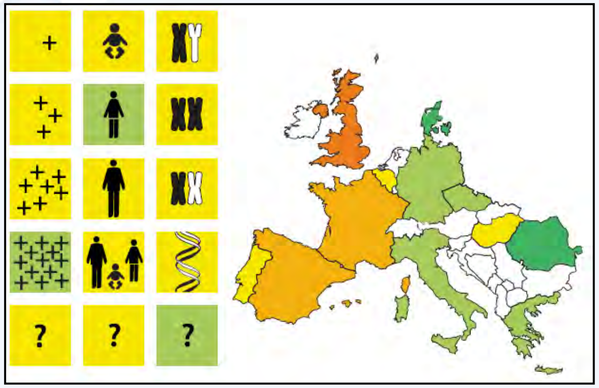
If a patient representative is a member of the register's governing board, indicate the importance of his/her opinion according to the domains of concern . (Multiple choices, expressed as score)



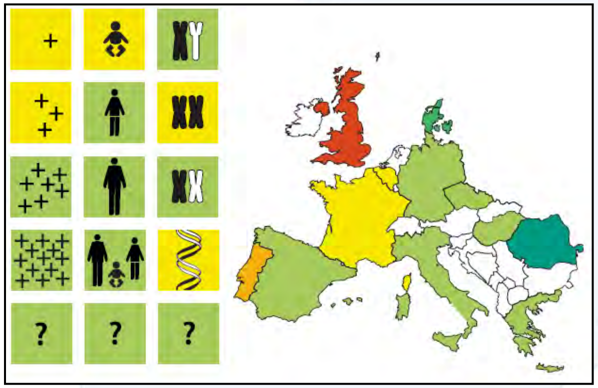
76% Registry aims



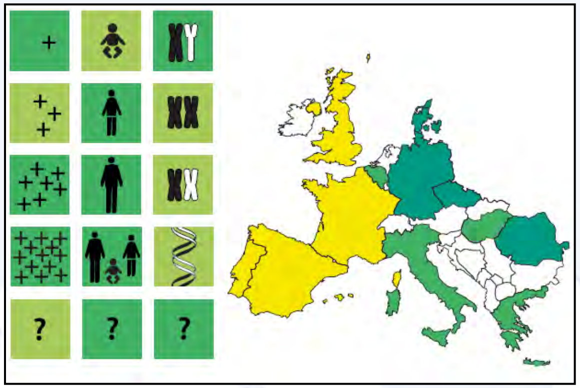
74% Ethical and legal issues



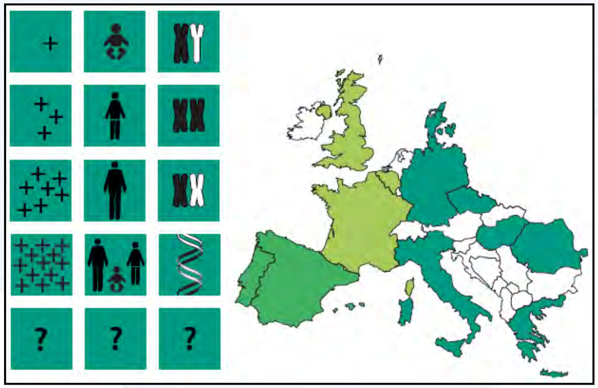
67% Access to data



64% Communication with users



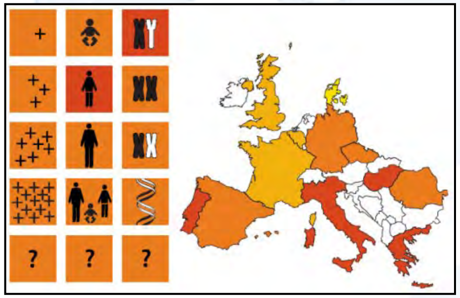
60% Stakeholder alignment



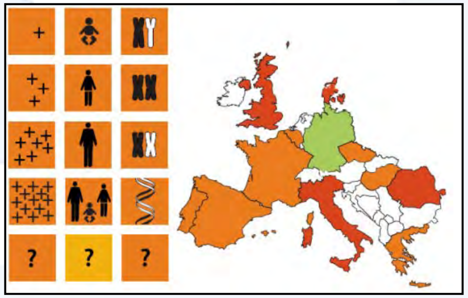
51% Financial and administrative issues

Long-term Financial Sustainability

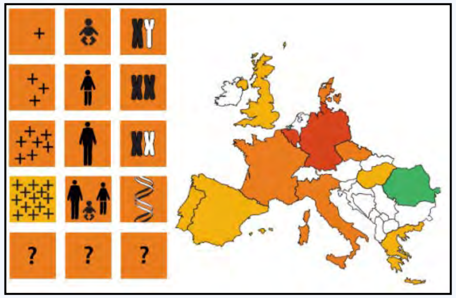
The value of a register is measured by its longevity. Among the following funding sources, indicate the 3 that could best assure the long term financial sustainability of the register. (Multiple choices, expressed as score)



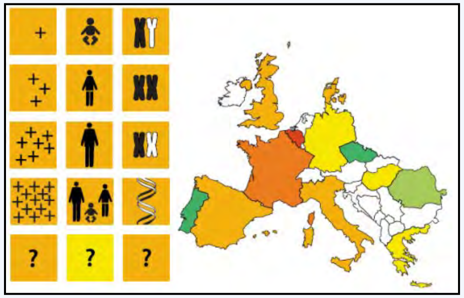
42% Patient organisation



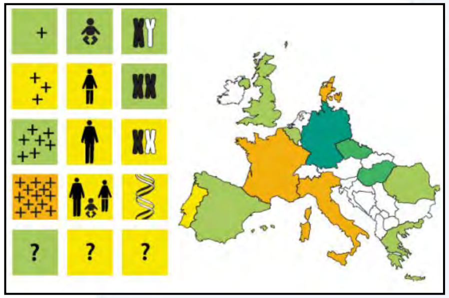
40% National authority



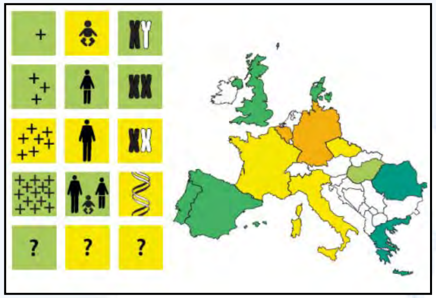
39% University/research institute



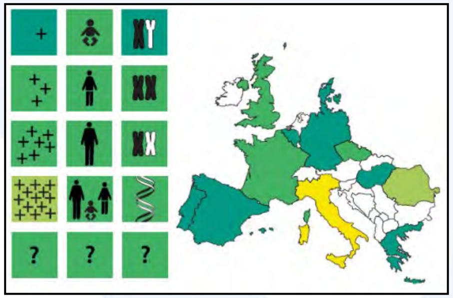
28% Patient organisation



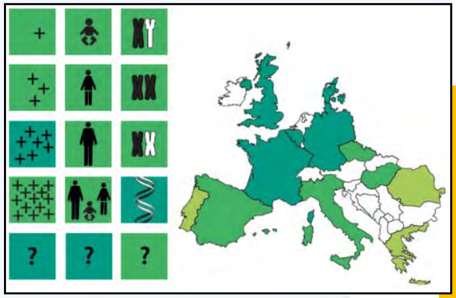
20% Hospital



19% Foundation



12% Regional authority



11% Industry

European Platform for Rare Disease Registries

European Framework and Infrastructure

- Overall, the overwhelming majority of respondents (84.8%) reported being in favour of a uniform legislative framework for RDPR across Europe, where only 4.3% did not agree and only 10.9% had no opinion.
- An overwhelming majority of respondents (90.7%) agreed with a common European registry infrastructure. Only 2.8% disagreed and 6.5% had no opinion regarding this proposal.

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Specific Results

- Some variability emerged for preferences regarding structural elements disease across groups and disease characteristics that represent distinct needs.
- Little or no variablity across disease groups for other registry elements
- Little or no variability across countries.

Policy Impact

- The policy impact of these findings suggests that national preferences and disease-specific preferences can sometimes be addressed by a common European registry infrastructure. For other preferences, disease specific, national or regional initiatives may be more appropriate.

Ethical and Social Implications

- The EU Charter of Fundamental Rights outlines patient rights to privacy of sensitive data, the right to participate freely in research and to contribute data in the name of solidarity. But it also recognizes the right of access to preventive health care and benefit from medical treatment.
- This strong overall consensus around the structure and uses of patient data illustrates the need for a careful balance in patient rights and societal “duties” in research participation.
- It is critical that any activity in patient registration and data collection respects the needs and expectations of individual participants

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RD Connect

- Clinical data does not provide the full picture
- 80% of rare diseases are genetic and thus genetic sequencing (determining the precise order of nucleotides within a single gene, set of genes or entire genome) hold great promise for potential gene-based treatments.
- Additional biosamples may needed for further research

RD Connect

- Currently clinical data (registries), genetic data and biorepositories exist separately
- The RD Connect project aims at integrating these data sources and adding value to the data by developing analytical tools to better understand disease mechanisms, ultimately leading to improved diagnostic capabilities and new potential therapies.
- Additional ethical, legal and social considerations