RD Connect

WP 6 ELSI & PATIENT

IRDiRC INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

SEVENTH FRAMEWORK PROGRAMME

The need of unique identifiers

- The risk of re-identification versus the need to identify individuals – ethical aspects of patient unique identifiers in rare disease research

- Hansson Mats G (1), Lochmüller Hanns (2), Riess Olaf (3), Schaefer Franz (4), Orth Michael (5), Rubinstein Yaffa (6), Molster Caron (7), Dawkins Hugh JS (7,8,9,10), Taruscio Domenica (11), Posada Manuel (12), Woods Simon (13)

Code of Practice

- Code of Practice for regulated access to data – A report of an ethical and legal framework for sharing and access to data in Rare Disease Research
- Mats G. Hansson, Jane Reichel, Sergi Beltran, Ivo Gut. Rachel Thompson, Olaf Riess, Franz Schaefer, Hanns Lochmüller,
- See the RD-Connect website!

Revised Ethical framework 1

- The revised ethical frameworks in RD Connect complements and build on the first stage ethical framework in which data and Sample sharing was regulated in a Charter. (2015 EJHG) as the result of a careful negotiation of different stakeholders' interest, now also endorsed by international organizations such as IRDIRC, BBMRI.SE and BBMRI.IT.
- With the same participatory procedure, a new paper has been published in the EJHG. The statement paper, provides guidelines for consent in Rare Diseases Research and builds on current literature analysis and on the results of empirical work carried in collaboration with the partners in RD-Connect (EJHG 2016); Patients Groups were directly involved in the drafting phase .

Children Involvement in Longitudinal research: Report from workshop

- Paediatric biomedical research within the field of Rare diseases (RD) is crucial if we are to gain insight into early onset pathologies, detect carrier status and develop new therapies.
- There is increasing agreement on the need for children to be part of the consent process, but assent should not be an absolute requirement
- Certain contents should be conveyed depending to age maturity when the child or the young can make sense of the information.

Children Involvement in Longitudinal research: From assent to Consent 2

- Requesting assent when the child does not understand the implications only means to abuse it in order to get an official OK for research. Children who have donated samples to biobank research should be contacted to legally consent when they reach majority.
- The practical side for re-contact could easily be solved using already existing IT-solutions where contact information can be kept updated (for example using a system like the Rudy study).
- For already existing collections, offering participants the possibility to opt out should be the minimal requirement. Ethical review boards may issue ethical waivers.

Communication of pre-manifesting Carrier Status: main recommendations

- Pre-manifesting carrier status is an issue that researchers and doctors working with rare disease patients have to face.
- Further research is needed in order to assess stakeholders attitudes and preferences around family sharing.
- Individuals and families involved in research should be aware of the possibility that pre-manifesting carrier status may be an outcome of the process and may involve relatives. The development of an ad hoc policy for communication and related information strategies is recommended.

Genetic data in public research databases: Which governance mechanisms should apply?

International meeting open to stakeholders. WP6 educational training (27/28 April 2016) in collaboration with CHIP ME (EU cost action)

Aims:

- to explore ethical and legal challenges that may arise when researchers are required to deposit genetic and genomic research data in public research databases
- investigate governance mechanisms that may support ethically and legally compliant data deposit

EJHG Open

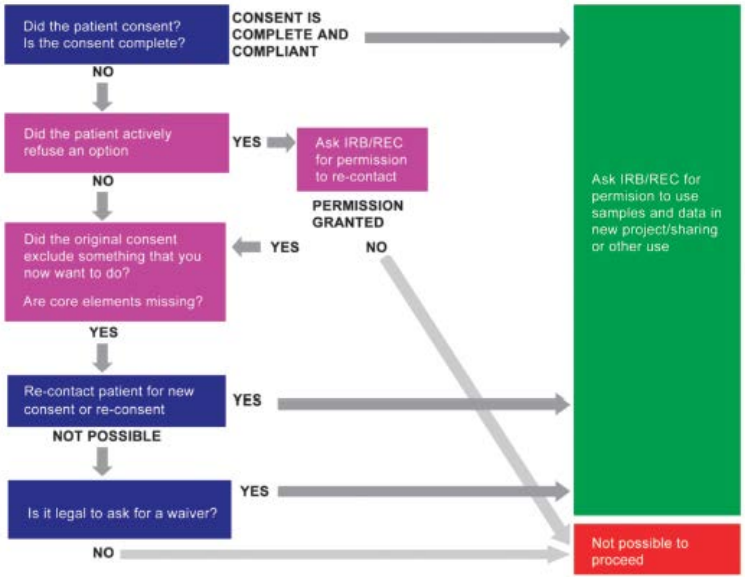
European Journal of Human Genetics (2016), 1–7

www.nature.com/ejhg

ARTICLE

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

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



Box 1 Essential elements of IC documents for International Consortia on RD

Information elements that are relevant in IC documents for biobank and observational studies in RD Research

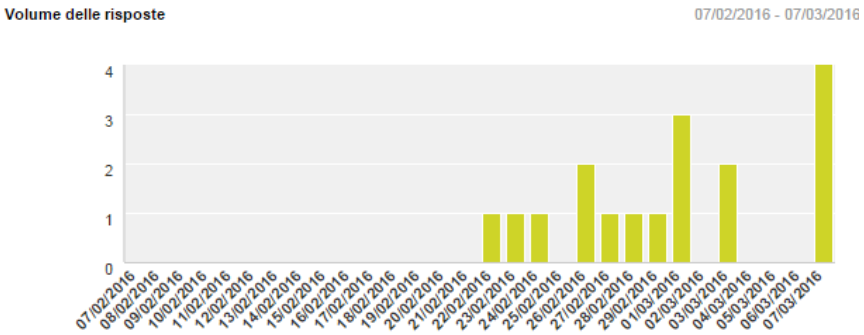
- General (name of the PI, Institution, funding, duration, oversight, contact persons)
- Aims, research uses of data (eg, cancer research and RD research)
- Voluntariness of participation and possibility to withdraw
- Procedures involved in participation, including interviews and blood taking
- Kinds of samples and data that will be collected
- Potential physical, psychological and social risks (informational risks)
- Potential benefits of participation
- Protections in place locally to ensure the confidentiality of samples and data
- Access to data/samples for research purposes: who will have access, who should control and what the procedures in place (data access committee)
- Access to data/samples for purposes such as validation and quality control
- Study oversight
- Compensation/reimbursement
- Custodianship of samples
- Study dissemination plans (professional journals/lay versions/codified or aggregated results only/specific results/patient pictures and occasionally short professional video sequences).

CEs for the IC of studies participating to international RD research

- Possibility of data sharing across research groups and national borders
- Possibility of large-scale genome sequencing techniques
- Return of secondary findings
- Hosting of the data in open access database (eg, in RD-Connect the European Genome-Phenome Archive)
- Use of interoperable identifiers for the de-identification of participants
- Access by industry if foreseen and prospects for third-party commercialisation and intellectual property
- Possible linkage to different data (registries, medical records, etc.)
- Withdrawal procedures, such as sample retrieval and/or destruction and difficulties in ensuring the right to withdraw for the data already shared
- Permission to re-contact

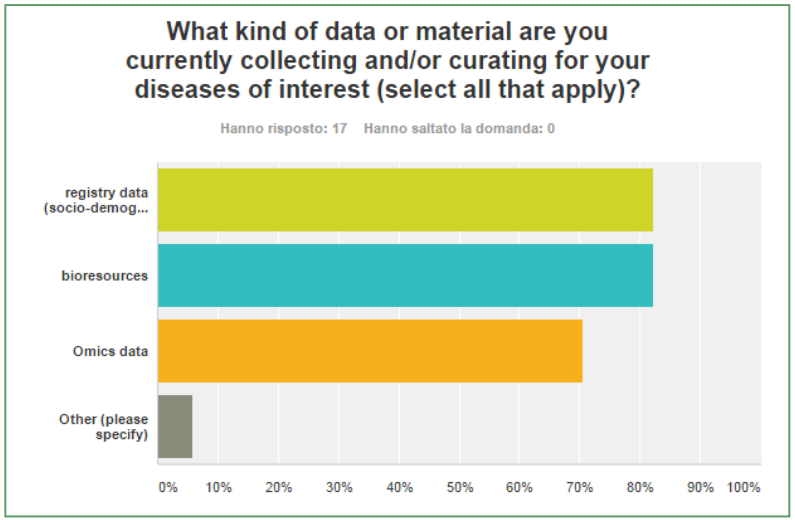
RD Connect

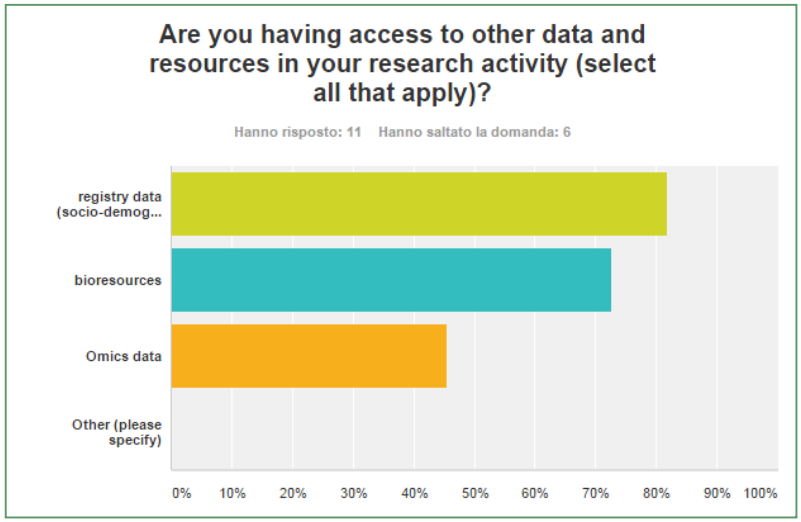
EURenOmics survey on informed consent and re-consent (n=12/37)



France (4)
UK (3)
Netherlands (2)
Belgium(1)
Italy (1)

Just a quick overview





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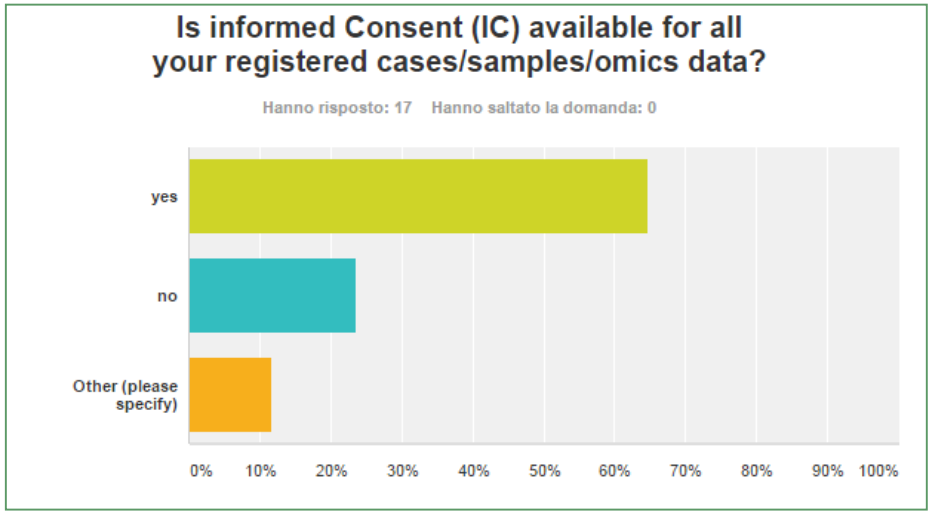
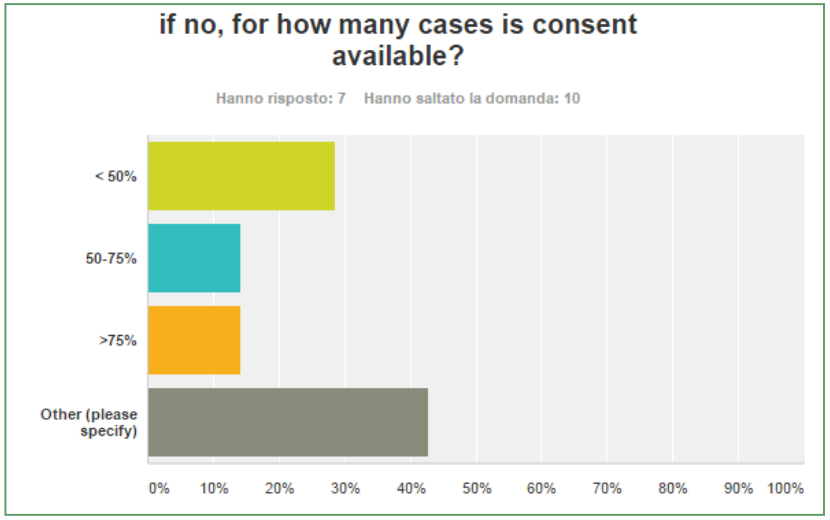


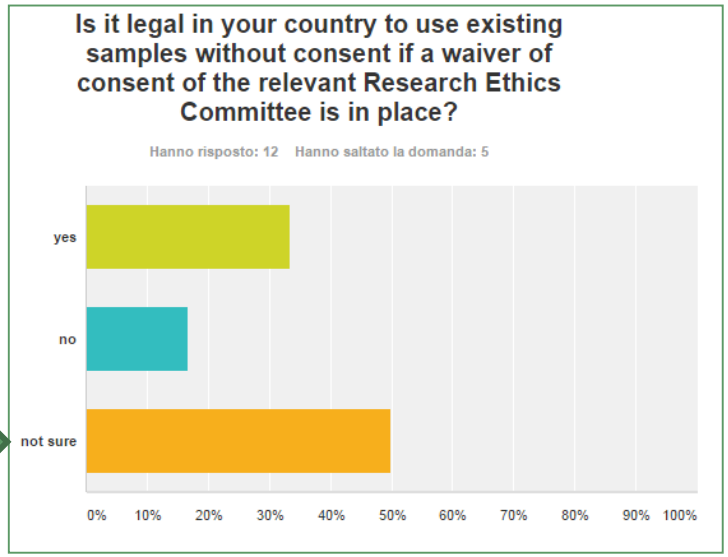


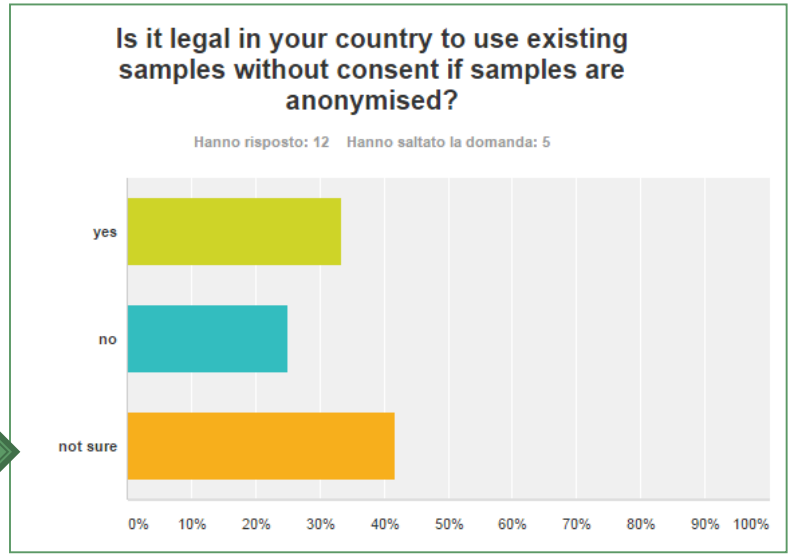
depending on the data; for blood > 80%, for DNA 100%, for clinical data < 50%

I cannot answer this question since it depends of the type of consent you mention: if it is consent for genetic testing we have consents in approximately 90% of cases, for data sharing it is below 10%

…the most ethically acceptable solutions where re-consent is impossible

Opzioni di risposta	Risposte	
A waiver of consent by the Research Ethics Committee (REC) or Institutional Review Board (IRB)	84,62%	11
Personal notification with the possibility to opt out (presumed consent unless the participant actively requests cancellation)	15,38%	2
Wide dissemination of the information through the work of patients associations and moral endorsement of these	15,38%	2
Anonymisation of data and/or samples	61,54%	8
Other (specify)	15,38%	2
Totale rispondenti: 13		





"You should at least ask"

- "You should at least ask": The expectations, hopes and fears of rare disease patients on large scale data and biomaterial sharing for genomics research - *EJHG in press*
- This work is *unique* in exploring the views of people with a range of rare disorders from many different countries. Key findings
  - Risk/benefit assessment by patients/participants is nuanced
  - RD patient advocates are not more permissive with data than those with CD or general population
  - But blurring of research/clinic divide can mean participation is not always a free choice
  - Vulnerability and discrimination
- UNEW-PEALS (WP6) were also instrumental in the Delphi set up and methodology which is covered in WP8 achievements

WP6 Last Publications

- Isabelle Budin-Ljøsne, Deborah Mascalzoni, Sirpa Soini, Helena Machado, Jane Kaye, Heidi Beate Bentzen, Emmanuelle Rial-Sebbag, Flavio D'Abramo, Michał Witt, Geneviève Schamps,Višnja Katić, Dusanca Krajnovic, Jennifer R. Harris, "**Feedback of individual genetic results to research participants: Is it feasible in Europe?**", BMC Medical Ethics, (in press) 2016
- Pauline McCormack, Anna Kole, Sabina Gainotti, Deborah Mascalzoni, Caron Molster, Hanns Lochmuller, Simon Woods "**You should at least ask" The expectations, hopes and fears of rare disease patients on large scale data and biomaterial sharing for genomics research"**. EJHG 2016
- Sabina Gainotti, Cathy Turner, Simon Woods, Anna Kole, Pauline McCormack, Hanns Lochmüller, Olaf Riess, Volker Straub , Manuel Posada, Domenica Taruscio, Deborah Mascalzoni, "**Improving the informed consent process in international collaborative rare disease research: effective consent for effective research"**, EJHG 2016
- Isabelle Budin-Ljøsne, Harriet Teare, Jane Kaye, Deborah Mascalzoni, "**Meta consent: Is it new and is it fit for purpose?**" BMJ, 04. Nov 2015, Http://www.bmj.com/content/350/bmj.h2146/rr-0

Acknowledgements/collaborations

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SEVENTH FRAMEWORK
PROGRAMME

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