RD Connect: Big data for rare disease
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Presented by Genetic Disorders UK
in partnership with Global Genes – Allies in Rare Disease
#GDLS2016
BIG DATA FOR RARE DISEASE

Pauline McCormack, Newcastle University
Recent years have seen an increased focus on RD research, with funding opportunities from different areas.
But: risk of data silos increases
Sharing: What?

- Raw data from all types of studies
- Genomic data
- Phenotypic data
- Natural history data
- Clinical trial data
- Biosamples (blood, DNA, tissue samples, cell lines…)
- Access to participants
- ...

RDConnect
Sharing: Barriers

- **General**
  - Privacy protection issues: “do I have the patient’s permission?”
  - Lack of infrastructure: “I want to share data but where do I put it?”
  - Lack of standards and interoperability

- **Academia & Industry**
  - Culture of protecting research results
  - Lack of incentives for sharing
  - IP issues/competition
Sharing: Benefits

- Overcoming the “rare disease problem”
  - Cohort size
  - Powering trials
  - Finding confirmatory cases
- Reducing costs
- Reducing duplication of effort
- Facilitating validation of results
- Enabling engagement with experts and the patient community
An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research

Overarching objectives:

- Contribution to the IRDiRC objectives of delivering 200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020
- Development of an integrated, quality-assured and comprehensive platform in which complete clinical profiles are combined with -omics data and sample availability for rare disease research.
RD Connect

- €12m euro EC contribution, 6 years [2013-2018]
- Co-ordinated by Institute for Genetic Medicine, Newcastle University, UK
- Working alongside EURenOmics and Neuromics – genome sequencing for rare renal and rare neurological disorders (€ 12m each)
- Support IRDiRC – communications/organisation (€ 2m)
- €40m total
- Part of Global Alliance for Genomics and Health
RD-Connect partners

29 full partners, 26 associated partners, 20 countries
Patient Advocates Included in Decisions about the Platform

- Inclusion of patient advocates in decisions about the platform
- 16 member Patient Advisory Council overseen by EURORDIS
- Patient and Ethics Council (RD-PEC), examines social, ethical and legal aspects

Scope Issues around Data Sharing, Linkage, Consent, Children’s Inclusion

Explore with rare disease patients and patient organisations their hopes, expectations and concerns regarding genomics research
Publications on participant issues

- “You should at least ask”. The expectations, hopes and fears of rare disease patients on large scale data and biomaterial sharing for genomics research (*in press*)
- Improving the informed consent process in international collaborative rare disease research: effective consent for effective research (*in press*)
- International Charter of principles for sharing bio-specimens and data
The importance of participation

- Focus for dynamic dialogue between researchers and patients
- Inter-understanding increases uptake and acceptance of technologies
- Researchers know developments are relevant and needed
- Promotes high professional standards and ethical integrity
- Builds trust and confidence around new developments and technologies
The importance of participation

- All participants have given consent for international data sharing
- No personally identifiable information
- Researchers will be checked before being given access
- All usage will be tracked
Data example - biobanking

- Cataloguing and registration of rare disease biobanks
  - Biobanks can sign up and give details of their biobank in an “ID card”
  - Allows biobanks to participate in RD-Connect infrastructure and research
  - Standardised assessment procedure, MTAs etc.

- Sharing sample-level data in a common database
  - Not just sample numbers but drill-down right to individual samples
    - Researchers can find the samples they need for their research
    - Allows data be traced back to the sample it came from for further research
RD-Connect Platform

Genomics

Samples selection

Variant Type

Genotype Feature

Variant Class

Results 2

Samples
Data in the RD-Connect platform

More exomes new projects

NCNP Japan 500 exomes

CMG Slovenia 300 exomes

NeurOmics 1,000 exomes

EURENOMICS 1,000 exomes

SeqNMD (Broad) 500 exomes

MyoSeq (NCL) 1,000 exomes

CNAG Rare 300 exomes
Matchmaker Exchange
How can you be involved?

- RD patient advocate
  - Fill out an ID Card to make your registry/biobank visible
  - Get in touch if you would like to work on development
  - Tell researchers/clinicians you are working with about the system
  - Sign up for the newsletter

- Researchers – share your WES/WGA data

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Some concluding thoughts

- Collaboration and data and sample sharing are crucial in rare disease
- Opportunities arising from the increase in RD research will be missed if projects do not share data and samples
- Number of projects that have been made possible as a result of sharing are proof that hurdles can be overcome
- Requires a change in mindset on all sides
- Requires appropriate infrastructures
- Requires preparation to share data (consent needs to cover sharing)
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