Experiences with building and managing a registry

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Ring14 International
The Italian Ring14 Association was a non-profit organization founded in 2002 by a group of families with children affected by rare genetic diseases caused by molecular rearrangement of chromosome 14.

In some cases, one of the two chromosomes assumes the form of a silent ring, in others, the pair of chromosomes loses or transfers genetic material. The consequences of these alterations are really severe.

The Ring14 syndrome
- Very rare disorder (prevalence: <1:1,000,000 Orphanet)
- Omim #616606 (obtained October 2015)
- Currently there are no effective therapies for the Ring14 syndrome
Ring14 Syndrome: SYMPTOMS

- EPILEPSY
- RESPIRATORY INFECTIONS
- MOTOR DELAY
- LANGUAGE DELAY
- AUTISTIC TRAITS
- MALFORMATIONS (heart, kidney, retina)
- FEEDING PROBLEMS (malnutrition, dehydration, anorexia, celiac disease)
- RARE DISEASE
Ring14 International, founded in 2014, is a non-profit worldwide organization aimed at networking the single national units which share same characteristics and purposes.

Ring14 International aims is to manage clinical assistance programs (to “care”) for families and promote scientific research projects (to find a “cure”) at a centralized level.

Ref.: Azzali et al, J Genet Disor Genet Rep 2015
# Projects promoted by Ring14 International

- **BIO BANK**
- **CLINICAL DATA BASE**
- **INTERNATIONAL CALLS FOR RESEARCH PROJECTS (2016 call awards in Nov.)**
- **ORGANISATION OF INTERNATIONAL WORKSHOPS**
- **ACTIVITIES FOR FAMILIES (international meetings, Guidelines for assistance)**
Why we need a clinical registry? Aims...

Rare diseases are highly heterogeneous

Registries bring together a (small) patient population, which can be used to:

- Improve the quality of life of patients
- Complete the natural history of the disease
- Engage academia and pharma companies

1) Registries allow clinicians to apply the best care path

2) Registries measure patient progress and follow-up outcomes

3) Provide disease knowledge which makes the disease easier to be studied, increasing the probability a treatment can be developed in the future
The «ideal» patient registry of Rare Diseases...a general management system for different stakeholders

Patient-centric
- optimized for a disease
- Specificity for geographic areas (population-based registries)

Tool system for clinicians
- Integrate with hospital based labs, Rx department and IT system
- Longitudinal records

Service for the scientific community:
- Genetic research
- Engage university
- Clinical trial studies
Ring14 clinical database: features

Enabled clinician/scientist users: 9 (have to sign a disclosure)

# of profiled patients: 61 (~30 to be recorded in the next months)

Data collected:
- Personal data
- Family history/pregnancy
- Genetics
- Growth & development
- Neuropsychiatry/neurology/Comorbidities
- Common form to collect data

Interoperability

Statistics/Reports

Achievements: 7 published papers (4 in preparation), 1 awarded grant by Telethon (plus another one recently submitted to NIH)

Organised by Istituto Superiore di Sanità
Rome (Italy), September 26-28, 2016
Management of a Clinical database for RDs: issues

- $$$/sustainability (Ring14 invested €30K 2006-to date)
- “Internationality” perspective
  - Translation
  - Clinical data harmonization
  - Easy accessibility/usability
  - Paucity of common procedures & standards
- Privacy
  - EU regulation on data protection (679/2016)
  - Informed Consent
  - Access with dual-password authentication
- Interoperability measures
  - Linking biomedical data across different platform (biobanks & academy)
  - Globally Unique IDentifier
- Dedicated staff
  - Data entry managed by a MD
  - IT manager
Thanks to ...

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Partners: