BRIDGEHEALTH

BRidging Information and Data Generation
for Evidence-based Health Policy and Research

Il Registro Nazionale delle Malattie Rare (RNMR)

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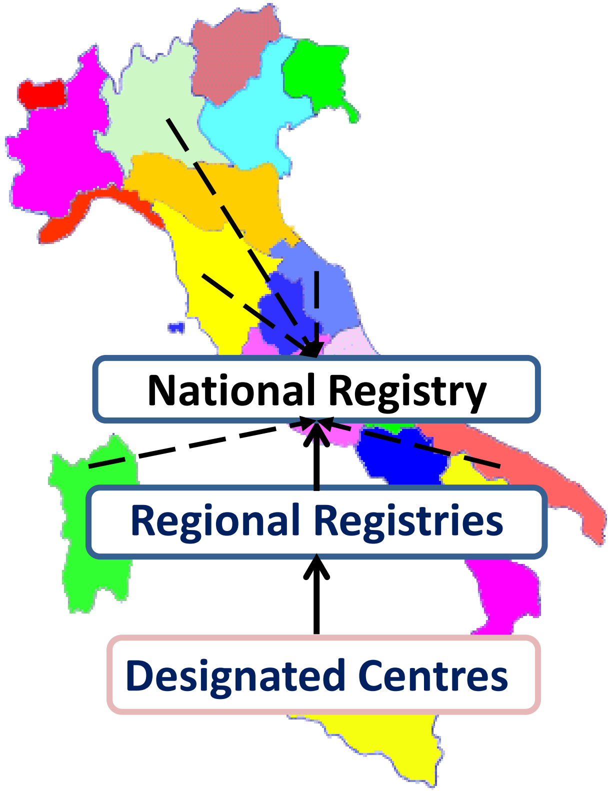
Features of RNMR

- Registry of rare disease (RD) patients
- Mandated by law (Ministerial Decree 279/2001)
- Functionally connected to RD Regional Registries (RR)
- RR data sources: Centres designated to ensure RD care and assess the right to special rules of cost exemption

- Aims: RD surveillance, service planning, research



National Registry

Regional Registries

Designated Centres

Pros and Cons

- Stability
- Full national coverage
- Patients streamlined to selected centres

=> ≈ population based

- No need for consent
- 500-2000 diseases covered

- Non-vocational => Sensitivity, minimum data
- Stiff => Difficult to adapt to new knowledge

Developments

- Combine data sources:
  - EHR
  - Mortality registries
- Centralize clinical research registry management:
  - Paroxysmal Nocturnal Hemoglobinuria database
  - Lesch-Nyhan disease registry
  - Combined immunodeficiencies registry
  - ....
- Extend data set and aims based on international platforms
  - EPIRARE: public health focus; BoD, disability profile; QALYs
  - RD-Connect: focused on research, biobanks and «–omics» platforms