FAIR genomes: a national guideline to promote optimal (re)use of NGS data in research and healthcare

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FAIR genomes: a national guideline to promote optimal (re)use of NGS data in research and healthcare

- Promote large scale (re)use of all human genomic data in the Netherlands to maximize knowledge extraction for research and healthcare
- By harmonization of essential aspects of the genomics data workflows and reporting across all data-generating centers in the Netherlands (germline rare disease, common disease and oncological data applications)
- While considering needs of all stakeholder groups (e.g. patient/participant, laboratory staff, clinicians, bioinformaticians, researchers)

- **Scope:** NGS (meta)data. **Out of scope** (part of other projects): ELSI, security, linkage, ...
FAIR?

- Easy to find and understand
- Well described conditions for data access
- For data to be combined
- For future (re)use in care/research and replication purposes

FAIR and resources from https://www.ands.org.au/working-with-data/fairdata/training
The route towards a national guideline to promote optimal (re)use of NGS data in research and healthcare

Implementation
Dissemination
Training

large scale (re)use
Approved Traceable Access
Benefits all stakeholders: Patient, Researcher, Physician, etc.
Implementation Dissemination Training
Some of the current challenges in patient care for Dutch genome diagnostic laboratories/community

• Exome analyses in routine diagnostics -> Interpretation huge challenge

• Sharing/exchange of clinical information
  • Information on individual DNA variants is often absent in public databases

• Comparable (harmonised) results/reports between laboratories

• Privacy issues/informed consent
  • Different rules and regulations
Ambitions to improve patient care by Dutch genome diagnostic laboratories

- Share knowledge about DNA variants
  - Sharing reduces time for interpretation
- Platform for harmonisation discussion
- Not only research also health care needs to become FAIR!
- Enable finding of “rare” patients
Challenges and ambitions in research:

• FAIR genomes and FAIR clinical (meta)data together with proper context:
  • Ease collaboration (especially on (inter)national scale)
  • Make data driven medicine/Artificial Intelligence (AI) possible (AI context sensitive)
• ‘Solving’ more patients
  • Enable use of ‘all data collected’ for N=1
  • Enable benchmarking
• Inform personalized approaches
  • Better treatment & prevention
• Health research
  • E.g. 1 million genomes (project) (including a catalogue of what data is available)
  • Paving the way for large research initiatives
1+MG project: a MS (funded) initiative
FAIRgenomes: (part of) Dutch contribution

Declaration for delivering cross-border access to genomic database

- 1 million genomes accessible in the EU by 2022
- Linking access to existing and future genomic database across the EU
- Providing a sufficient scale for new clinically impactful associations in research

BIOS: Biobank-based Integrative Omics Study
Planned deliverables:

What
• Guideline NGS data FAIRification
• Modular standards, considering differences in needs and processes
• Tools to ease implementation of the guideline
• Create critical mass/uptake in the professional groups via pilot projects

How
• Fit the existing Dutch situation minimizing cost and maximizing results
• Converge with many international developments
• National strategy with leading organisations driving implementation
## Results so far:

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<thead>
<tr>
<th>Stream2: Inventory of tools and infrastructure for genome data FAIRification</th>
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<tr>
<td>This is joint work between the FAIRgenomes project, the Netherlands X-omics Initiative and the United Against Metabolic Disorders (UAMD) project</td>
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### Semantic models and ontologies

- **For samples**
  - The experimental factor ontology (EFO)

- **For consents / data licenses**
  - ADA-M

- **For data use conditions**
  - Data Use Ontology (DUO)
To summarize:

FAIR genomes will help diagnostic labs to exchange results and address shared challenges (germline and somatic)

FAIR genomes is prerequisite for large scale multi-center analysis
- E.g. personalized medicine and data driven medicine (AI)

FAIR genomes is a broad national coordination action with international links

FAIR genomes is an open project
- Stakeholders: patients/citizens, health care, research, policy makers
- Contact us
Questions suggestions? Let us know!
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