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

Update WP1: Unifying workflow and manual to promote FAIRness of NGS data & 1+MG

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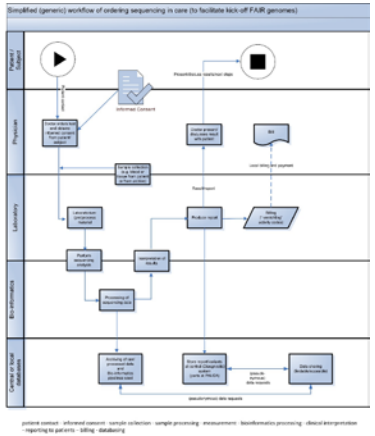
WP1:



Results so far:

- Work/process/data flow of NGS
- Contribution “Draft of Recommendations for WGS in genome diagnostics for rare disease”
- Best practices inventory: e.g. https://www.ga4gh.org/genomic-data-toolkit/
- Tools to ease implementation of (parts of) the guideline
  - iCRF generator (output from KWF TraIT2Health-RI project) : generate CRF(s) based on established and published codebooks @Nictiz
    - *All codebooks we produce: establish & publish @Nictiz?*
- 1+MG (co-lead of WG3 details later): contributions to (dynamic) roadmap (adopted yesterday)
- Participation in stream 1 & 2 FAIRgenomes
  - Results so far (clinical, sample, IC, ..): shared google document

WP1: next steps

Continue with unifying workflow and manual to promote FAIRness of NGS data

With subtopics (updates by):

- Guideline module informed consent: Daphne
- Guideline module (patient/subject to) sample to data: Joeri
- Guideline module from data to meaning: Peter-Bram+Gurnoor
- Guideline technical solutions/implementation(s): Jeroen Laros

1+MG:

- 1+MG is a EU-member state initiative (supported and financed by member states, incl NL)
  - ***1+MG initiative receives no direct EU-funding***
- 1+MG has 10 working groups
- I have, on behalf of NL, been asked to co-lead WG3 “Common standards and min. dataset for clinical and phenotypic data”
  - co-lead Augusto Rendon Genomics England (Brexit???)
- Each country has or is setting up a national mirror group (NMG;mimicking 1+MG structure)
- ZonMW funded:
  - FAIRgenomes
  - NMG-NL received start funding to establish NL-1+MG-actionplan

1+MG:

- Last November we (ELIXIR coordination & large group from 1+MG) applied for CSA grant: SC1-HCC-06-2020: Coordination and Support to better data and secure cross-border digital infrastructures building on European capacities for genomics and personalised medicine
- Results FAIRgenomes:
    - FAIRgenomes is input from NL in 1+MG
    - Relevant output of 1+MG also input for our guidelines
- NMG-NL writing NL-1+MG-actionplan (being reviewed)
    - Rick van Nuland & Gert-Jan van Ommen & Ruben Kok
- Roadmap 1+MG 2020-2022 has been adopted (Feb 4)
    - Commission will use roadmap to plug 1+MG-topic into new EU Horizon program

1+MG: participation NL

- Gert-Jan van Ommen & Roger Lim from VWS NL-representatives
- Ruben Kok WG1 (Scope, stakeholders and governance)
- Jasper Bovenberg & Susanne Rebers & Marjanka Smit WG2 (Ethical, Legal, and Societal Issues (ELSI))
- Jeroen WG3 (Common standards and min. dataset for clinical and phenotypic data)
- Edwin Cuppen WG4 (Good sequencing practice)
- Morris WG5 (Federated, secure, interoperable and privacy-respecting framework and access governance)
- Ilse Custers WG6 (Health economics and outcome research)
- Gijs Santen & Lisenka Vissers WG8 (Use case - Rare diseases)
- Edwin Cuppen WG9 (Use case - Cancer)
- Marian Beekman WG10 (Use case - Common and complex diseases)

BBMRI.nl Biobanking and BioMolecular resources Research Infrastructure The Netherlands

trait a Lygature platform

ZonMw

Nederlandse Vereniging voor Pathologie

Zilveren Kruis

health RI research infrastructure

KWF Kanker Bestrijding

elixir Netherlands

DTL Dutch Techcentre for Life Sciences

VKGL

Hartwig Medical Foundation

ELSI Servicedesk

VKGN

palga

VSOP

NFU Nederlandse Federatie van Universitair Medische Centra

Questions suggestions? Let us know!

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