

A JUNE 2020 UPDATE BY K. JOERI VAN DER VELDE



FAIR Genomes is a ZonMw "Personalized Medicine" project, nr. 846003201 **CONT**

Who are FAIR Genomes? WP leads



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</u>

from:

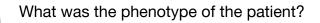
¹Radboud University Medical Center, Nijmegen, The Netherlands, ²University Medical Center Groningen, The Netherlands, ³University Medical Center Utrecht, The Netherlands, ⁴Amsterdam University Medical Centers, location VUmc, NL, ⁵VSOP - Dutch Patient Alliance for Rare and Genetic Diseases, ⁶Leiden University Medical Center, The Netherlands, ⁷Erasmus Medical Center, Rotterdam, The Netherlands, ⁸Durrer Center for Cardiovascular Research, Utrecht, The Netherlands, ⁹Maastricht University Medical Center, The Netherlands, ¹⁰Princess Máxima Center for Pediatric Oncology, Utrecht, The Netherlands, ¹¹Dutch Techcentre for Life Sciences, Utrecht, The Netherlands, ¹²Netherlands Cancer Institute, Amsterdam, The Netherlands, ¹³Hartwig Medical Foundation, Amsterdam, The Netherlands, ¹⁴Leiden Institute for Advanced Computer Science, Leiden University, Leiden, NL

Why do we need FAIR Genomes?



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





What kind of sharing is allowed by the consent?

Which tissue was sampled?

Which sample prep kit was used?



What type of NGS machine was used?

What software was used to perform read mapping?



Which protocol was used to interpret the data?

if FAIR:

"I would like to please have all nationally available VCF files for PBMC samples from cardiomyopathy patients sequenced whole-exome on HiSeq machines processed with GATK 4.0, for which consent allows re-analysis."

Now: not possible. *Let's make it possible.*



What is FAIR Genomes?



61 people from 14 institutes (NL). F2F meetings, Zoom calls, focus workshops.
Interacting with EJP-RD CDE, Solve-RD RD3, 1+MG, GA4GH, Phenopackets, X-omics, and others.

Currently **9 parts with 82 elements**: Personal (12), Clinical (16), Material (12), SamplePrep (7), Sequencing (7), Analysis (7), General Consent (8), Individual Consent (7), Study (6). **Together** we define what meta data is needed to **find, share and reuse** NGS data in research and healthcare. Forming an evolving **semantic** model for properties and values.

Focus on being **harmonized** with EJP-RD Common Data Elements, RD3, PhenoPackets, MIABIS, etc. Model, codebooks and applications all **free & open source software**.

Join us at: https://github.com/fairgenomes

Main result: a semantic model



Personal information

Meta-data element	Preferred ontology term for meta-data	Value types	lssue Number	Issue (open / closed)
Individual ID	NCIT:C164337	ID [string]	#3	closed
Gender	SIO:010029	Male SIO:010048 Female SIO:010048 Unknown / Undetermined	#4	closed
Genotypic sex	PATO:0020000	UNKNOWN_KARYOTYPE, XX, XY, XO, XXY, XXX, XXYY, XXXY, XXXX, XYY, OTHER_KARYOTYPE	#69	closed
Country of residence	SNOMEDCT:276205001	ISO 3166 country codes	#5	closed
Ethnicity	LNC:MTHU010275	ISO 3166 country codes	#6	closed
Country of Birth	SNOMEDCT:370159000	snomed ontological term [subclass of SNOMEDCT:370159000]	#7	closed
Year of birth (if allowed)	NCIT:C83164	YYYY [4 digits]	#8	closed
Inclusion status	SNOMEDCT:246097001	Alive / Dead / Lost in follow-up / Opted-out	#9	open
Age at death	NCIT:C135383	[Positive integer]	#10	closed
Inclusion criterion	OBI:0500027	Free text [string]	#11	closed
Primary affiliated institute	SIO:000688	List of Dutch institutes in BBMRI-ERIC	#60	open
Data available in other institutes	SIO:000688	List of Dutch institutes in BBMRI-ERIC	#62	open

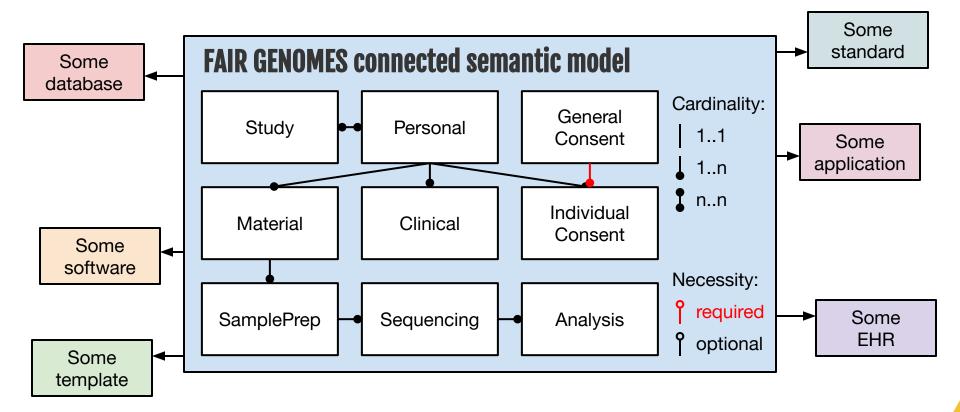
Note: both the elements and values are linked to **ontologies**.

All elements are compulsory, but with HL7 **NullFlavors** to tell why values are missing.

See:

https://github.com/fairgenomes/i nformation/tree/master/fairgeno mes_semantic_model

A model is a means, not a goal



UMCG Dept. of Genetics rijksuniversiteit groningen

Application #1: Nictiz

Gurnoor Singh, Jeroen Beliën, Sander de Ridder, K. Joeri van der Velde

are implementing a FAIR Genomes **ART-DECOR** codebook for

Nictiz (who develop & manage information standards for exchange of digital data in healthcare)



https://www.nictiz.nl/standaardisatie/art-decor/



https://art-decor.org/

See:

https://github.com/fairgenomes/information/tree/ master/fairgenomes_codebook_nictiz



Why an ART-DECOR codebook?

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For more useful tools and guidances, keep an eye soon have a Registry in a Box section!		health-ri.org), which will	

iCRF Generator is a Java program that can generate the core of an interoperable electronic case report form (iCRF) for several of the major electronic data capture systems (EDCs)

de Ridder S and Beliën JAM. The iCRF Generator: Generating interoperable electronic case report forms using online codebooks. F1000Research 2020, 9:81 (https://doi.org/10.12688/f1000research.21576.2)

Application #2: MOLGENIS

K. Joeri van der Velde, Gurnoor Singh, Fernanda de Andrade, Dieuwke Roelofs-Prins, Lennart F. Johansson, Bart Charbon & MOLGENIS Team

umcg Dept. of Genetics

MOLGENIS: scientific data platform **Powering ERNs** reate Subjects EUDID/ Research IF GENTURIS → with flexible model, tailor to your needs, 100+ instances running Ithaca Skin MOLGENIS.org If yes, specify fo Cranio fear of birth ndex status ' Solve-RD RD3 (https://github.com/molgenis/RD3_database) FAIR Genomes app Detailed NGS sample tracking https://github.com/fairgenomes/information/tree/master/fairgenomes molgenis app MOLDENIS Import data * Navigator Data Explorer Data Integration * Plagina * clinical codebook clinicaldiagnosis (#/FAR Geren

Public demo online



SFAIR Import data - Navigator Data Explorer Plugins -



VERSION 0.1 PUBLIC DEMO. THIS IS A PROTOTYPE FOR EVALUATION PURPOSES ONLY. DO NOT IMPORT ANY SENSITIVE DATA.

A national guideline to promote optimal (re)use of NGS data in research and healthcare.



Please visit & give us feedback!

https://molgenis93.gcc.rug.nl

FAIR Import data - Navigator Data Explorer Plugins -	Help Si
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Acral peeling skin syndrome	
Acroallosal syndrome	
Acrocardiofacial syndrome	
Acrocephalosyndactyly	

Bonus deliverable: new ontology

We will create a formal 'FairGenomes' ontology for important terms currently **not available** in ontologies

- → Genotypic sex (ie. karyotypes, ~10 items)
- → NGS kits (~625 items, source: BioCompare)
- Sequencing instruments (adding ~10 items)
- → Dutch hospitals (~110 items)

Future work



- Discuss open issues with FAIR Genomes & other domain experts
 Finalize attributes, descriptions, values, curate codebooks
 Further harmonize with other projects & existing standards
- We need to show that it works in *demonstrator projects*!
 Testdrive FAIR Genomes on genomics data (1+MG, B1MG, EJP-RD)
 See if works in daily healthcare & research practice (VKGN, KMBP, ..)
- Publish to share our hard work & FAIRify the rest of the world
- \rightarrow From meta-data to a real semantic data models ?
 - EMX format (MOLGENIS) works, but is not very semantic yet
 - Construct true semantic data model (RDF or JSON-LD)
 - Add rule based validator (RDF Shacl or JSON-LD markups)

Thank you.



Time for questions and anwers.