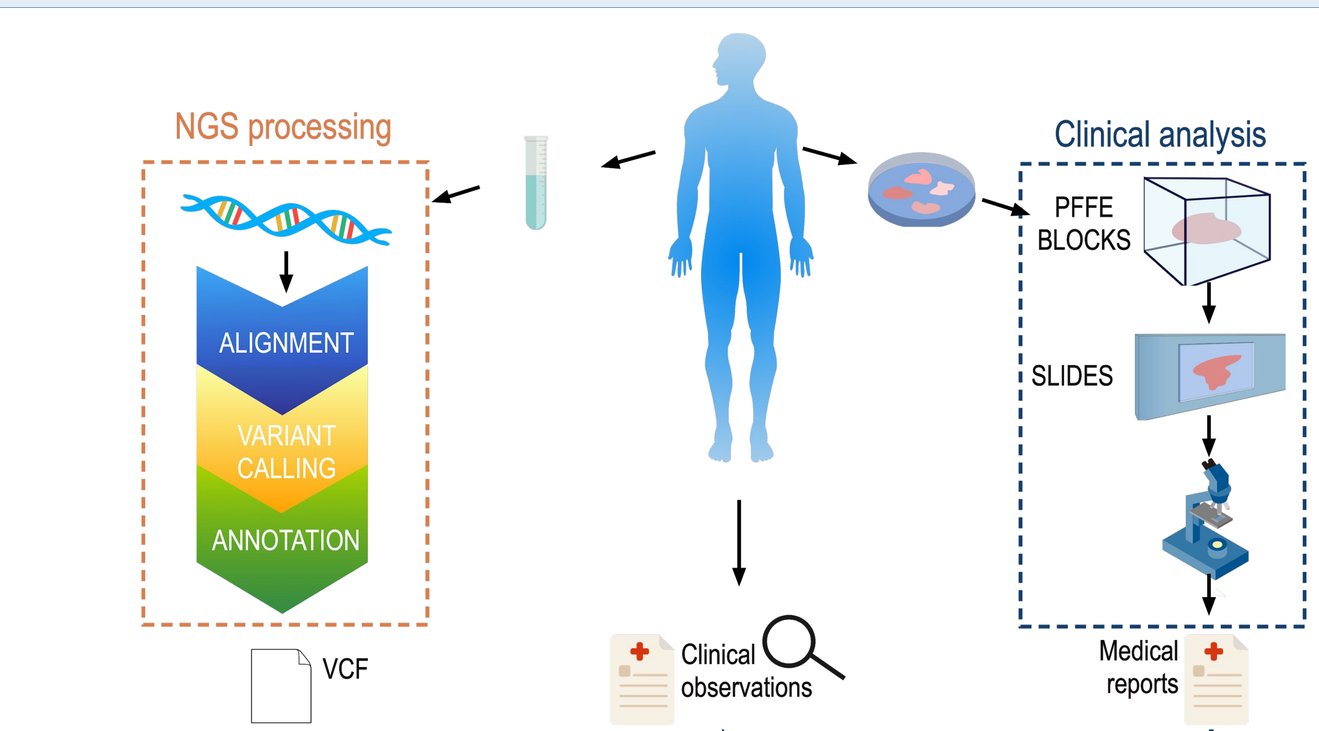
Open standards for semantic interoperability of clinical and genomic data: a proof-of-concept bridging openEHR, Phenopacket and FHIR

Francesca Frexia[1,2], Cecilia Mascia[1,2], Paolo Uva[1], Luca Lianas[1], Giovanni Delussu[1], Gianluigi Zanetti[1,2], Gideon Giacomelli[3,4], Diego Boscá[5], Silje Ljosland Bakke[6], Ian McNicoll[6], Heather Leslie[6].

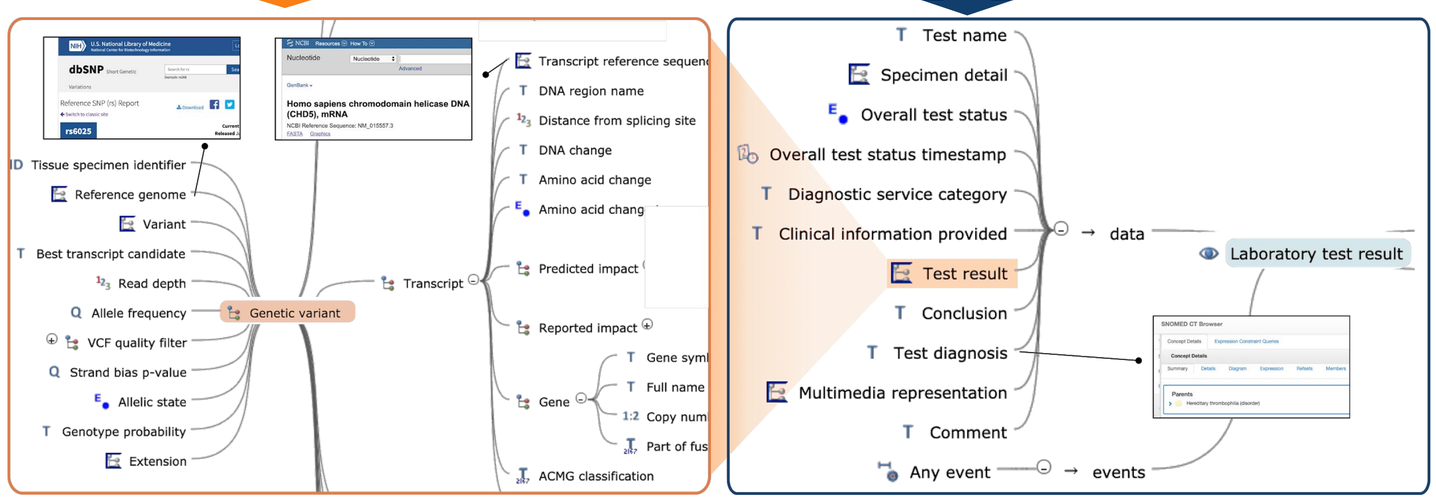
[1]CRS4: Center for Advanced Studies, Research and Development in Sardinia (Italy) - [2]European Joint Programme on Rare Diseases - [3]European-Canadian Cancer Network - [4]HiGHmed Consortium (Germany) - [5]VeraTech (Spain) - [6]OpenEHR Foundation

GA4GH 7th Plenary Meeting, October 21-23 2019

DATA GENERATION



DATA CAPTURE AND REPRESENTATION



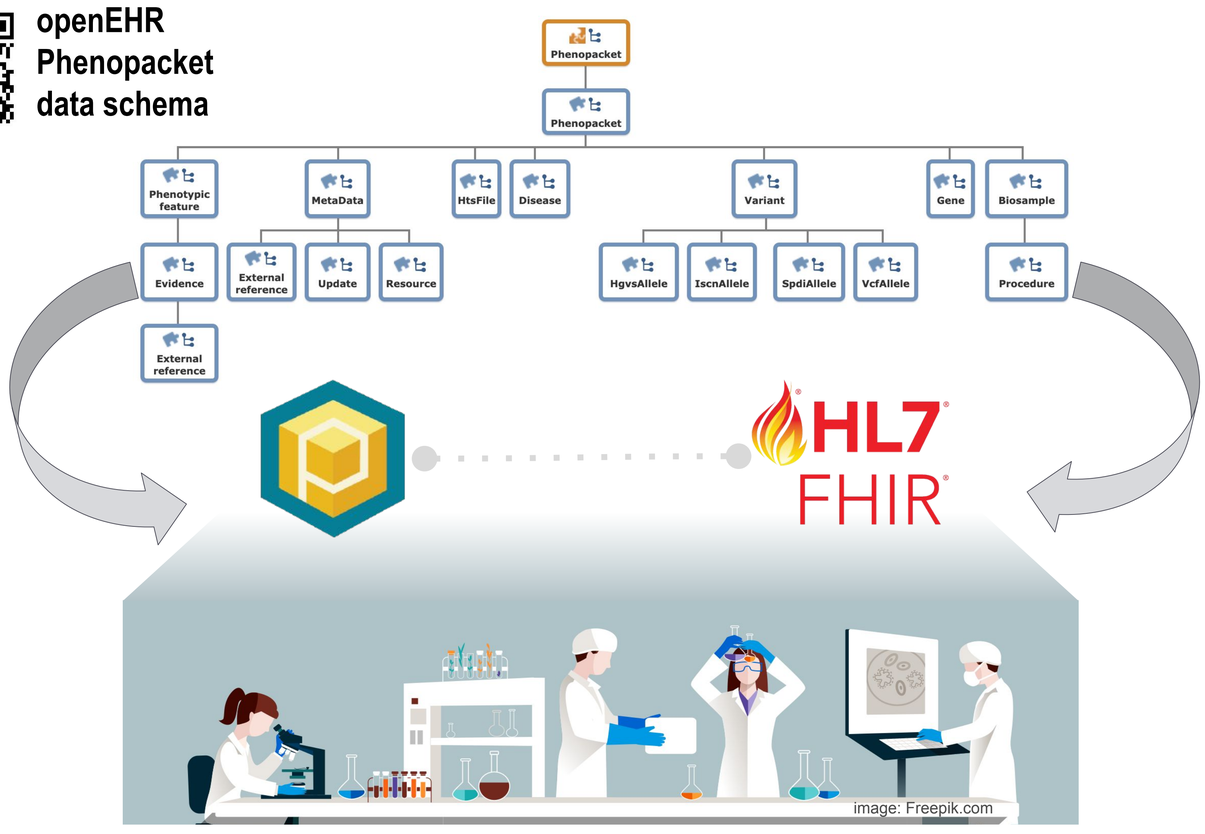
Genomic archetypes

*open*EHR

Clinical archetypes

DATA EXCHANGE AND INTEROPERABILITY

openEHR Phenopacket data schema



MOTIVATION

The quality of health information is strongly influenced by the ability of **capturing and maintaining data semantics** at the time of generation.

APPROACH

openEHR addresses these issues with:

- multi-level modelling, to collect clinical information with a computable and **ontological semantic approach**, regardless of specific software implementations;
- a wide community creating and managing a **broad library of clinical knowledge artefacts** (archetypes and templates), including models for **genomic information**.

RESULTS

- We implemented a system for the modelling, management and interexchange of biomedical data, leveraging **openEHR, Phenopacket** and **HL7-FHIR**.
- We represented the **Phenopacket data schema as openEHR archetypes** for the purposes of integration.
- We are testing the system with a synthetic dataset.
- Future works include the use with real datasets in the rare disease and cancer domains.

GA4GH Driver Projects: EU CAN. EUROPEAN JOINT PROGRAMME RARE DISEASES

Contact: francesca.frexia@crs4.it