Genomic Reporting Implementation Guide

source: 🔥 Home Page - Genomics Reporting Implementation Guide v4.0.0-ballot

Currently using the STU3 implementation guide. This acts as a standard for organisations to implement genomic reporting to FHIR.

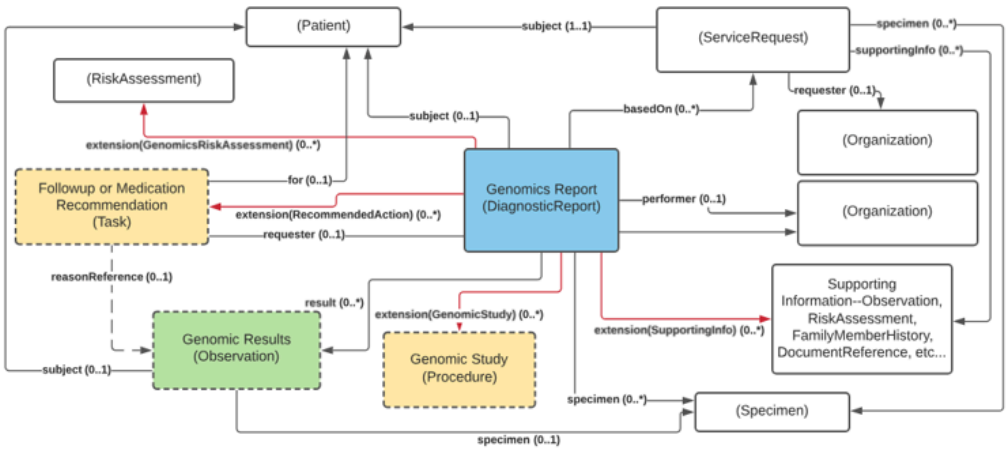
Main design principles

- minimise FHIR extensions
  - risk of data loss when passed to systems that do not support the extensions
- maximise reuse of resources = use of profiles
  - Observation
  - DiagnosticReport
- use separate observations for each independently useful observation(data).
  - This makes data discoverable and queryable
    - Links to evidence for supporting querying and the use of contained resources:
      - 🔥 Appendix D: Query Guidance - Genomics Reporting Implementation Guide v4.0.0-ballot
      - 🔥 References - FHIR v4.0.1

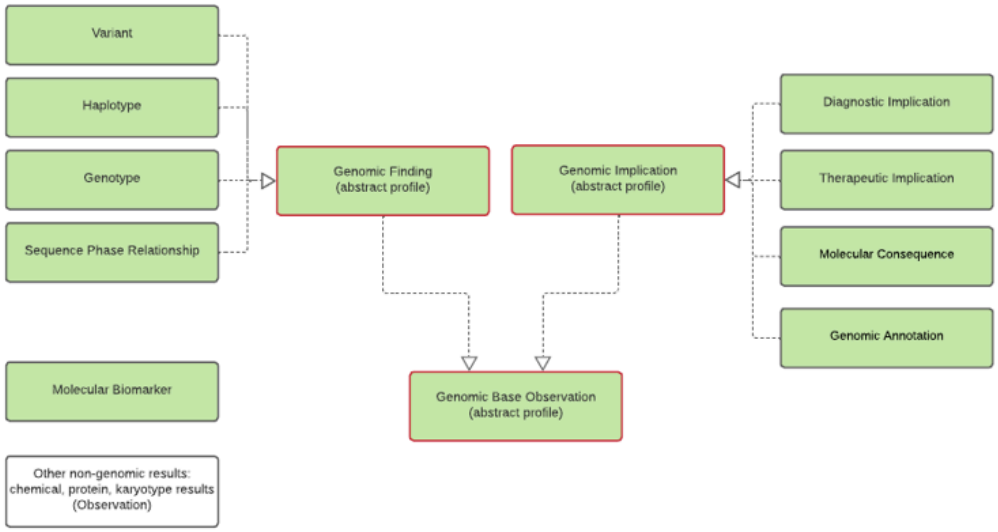
Documents for our FHIR design

🔥 General Genomic Reporting - Genomics Reporting Implementation Guide v4.0.0-ballot

- Basically we need to adhere to this design:



But what is Genomic Results? It is essentially a catchall term for many different Observation Profiles which all capture different data on our molecular reportables.
The example below shows all the profiles that extend from the Observation Resource.

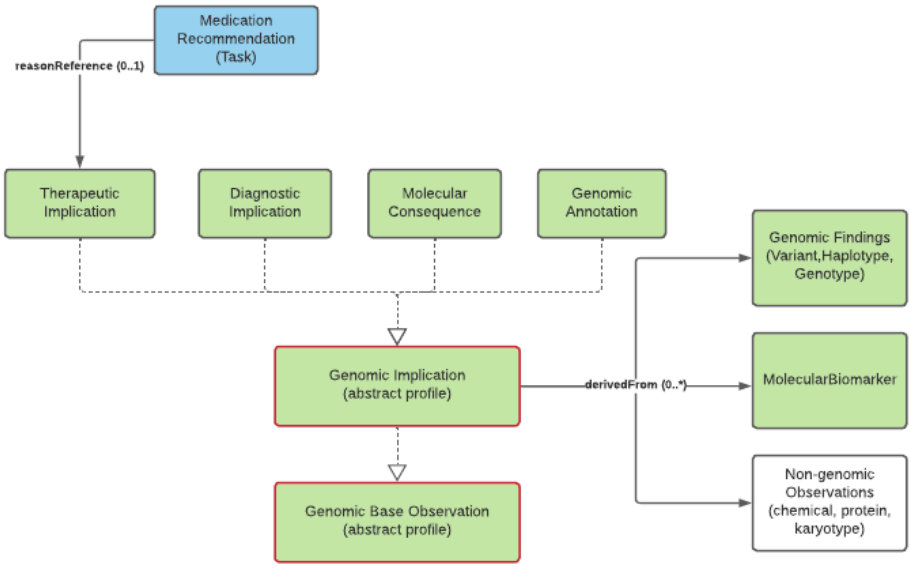


- Variant = stores all variant data

Everything else maps to Variant to indicate specific information about the variant

- genotype = used when a variant has genotype data
  - note the guide didn't state it, I just did it because it adheres to ZD db design
- diagnostic implication = variant used as evidence for a diagnosis
- molecular consequence = function impact of the variant
- genomic annotation = misc data that do not meet the above's descriptions

This is demonstrated in this diagram:



We can see that the Genomic Implication Profiles are derived from Genomic Findings = Variant/Genotype

Documents for querying molecular reportables

🔥 Appendix D: Query Guidance - Genomics Reporting Implementation Guide v4.0.0-ballot