Clinicalgenomics FHIR IG Proposal
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Genetic Reporting Implementation Guide

Owning work group name
Clinical Genomics

Committee Approval Date:
Please enter the date that the committee approved this IG proposal
• Feb 13, 2014

Contributing or Reviewing Work Groups
• Orders and Observations

FHIR Development Project Insight ID
1217

Scope of coverage
The Genetic Reporting Implementation Guide will identify and define extensions, profiles, terminology standards necessary for the reporting of clinical genetic/genomic/biomolecular findings and interpretations related to the domain of clinical genetics/genomics sufficient for precision medicine. This includes items like extensions, profiles, domain specific examples. This work will be defined using the available FHIR tooling and in accordance with documented quality guidelines. This includes, but not limited to, profiles on Observation, DiagnosticReport, ServiceRequest, Specimen, and Sequence.

At present, this implementation guide focuses solely on data structures - what data should be/might be present and how it should be organized. It does not address workflows around how reports are requested, created, approved, routed, delivered, amended, etc. The implementation guide is also paradigm-independent - the data structures presented here could be used in RESTful, messaging, documents or other mechanisms.

IG Purpose
Current Clinical Genomics FHIR work includes a number of profiles on Observation, DiagnosticReport, DiagnosticRequest, FamilyMemberHistory that relies on heavy use of extensions, and a Sequence Resource. Refactoring these profiles into a consistent approach, emphasizing components in the case of Observation, and separating them from FHIR core spec will simplify their use and understanding as well as conform to FHIR design going forward (reducing the number of Profiles in the core spec).

Content location
http://build.fhir.org/ig/HL7/genomics-unified/index.html

Proposed IG realm and code
uv/genreport

Short Description
This implementation guide tries to provide guidance that will enable improved interoperable and computable sharing of genetic testing results.

Long Description
Genomics is a rapidly evolving area of healthcare that involves complex data structures. There is significant value in sharing this information in a way that is consistent, computable and that can accommodate ongoing evolution of medical science and practice. The value comes from the ability to easily sort, filter and perform decision support on such information and the resulting improvements in care and reduction in costs such as the elimination of redundant testing. This implementation guide tries to provide guidance that will enable improved interoperable and computable sharing of genetic testing results.
This guide covers all aspects of genetic reporting, including:

- Human and veterinary as well as bacterial and viral specimens
- Representation of simple discrete variants, structural variants including copy number variants, complex variants as well as gross variations such as extra or missing chromosomes
- Representation of both known/catalogued variants as well as fully describing non-catalogued variations
- Germine and somatic variations
- Relevance of identified variations from the perspective of disease pathology, pharmacogenoomics, transplant suitability (e.g. HLA typing), etc.
- Full and partial DNA sequencing, including whole genome and exome studies
- Mosaicism (differing genetic characteristics for different specimens from the same subject)
- Mitochondrial DNA variations

Involved parties

- National Library of Medicine/NIH
- Boston Children's Hospital

Expected implementations

- National Marrow Donor Program
- Center for International Blood and Marrow Transplant Research

Content sources

No other source specifications, but we have gathered input from

- Global Alliance for Global Health (GA4GH)
- FDA
- National Academies
- ClinGen/ClinVar
- Variant Modelling Collaboration (VMC)
- CDISC PGx
- ISO TC215

Example Scenarios

- General Genomic Reporting
- Sequence Variants
- Cytogenetic Reporting
- Pharmacogenomic Reporting
- Somatic Genomic Reporting
- Genomic Profiling for Transplantation

IG Relationships

No, there not any IGs this resource depends on or that depend on this IG

Timelines

- May 2018 Ballot for R4
- Sep 2018 Ballot for R4 after ballot reconciliation

When IG Proposal Is Complete

When you have completed your proposal, please send an email to FMGcontact@HL7.org

FMG Notes