GenomeX - Genomics Data Exchange

The Problem

Genomic tests are usually sent from laboratories to health care organizations in the form of a PDF, isolating the data within the EHR and inhibiting its use for clinical care and research. This use case will support the collection of data in a standardized, machine-readable format, allowing this data to be used in many important ways.

Why this Use Case?

The HL7 Clinical Genomics Workgroup has been developing a FHIR Genomics Implementation Guide (IG), including an information model, definition of diverse genomic resources and guidance for the use and implementation of these FHIR resources for a variety of clinical genomic use cases. This in-depth work has taken place over many years.

It is time to accelerate the adoption of FHIR Genomics as a common, computable, data standard. Such adoption is essential to enabling the incorporation of Genomics into clinical care and the tremendous benefits which this provides. To do so, we must ensure that the FHIR Genomics Resources and Implementation Guide, meet the needs of stakeholders and that we gain expert feedback on potential areas for improvement and development of these resources.

The Solution
Working together - laboratories, EHR vendors and other interested stakeholders will leverage the work of the HL7/FHIR Clinical Genomics Working Group and their FHIR Genomic Reporting Implementation Guide, to design and build, scalable FHIR Genomics interfaces, therefore enabling appropriate data to be sent to EHRs or genomics repositories.

Expected Benefits

Standardizing the transmission of genomic data using Clinical Genomics FHIR Resources will increase the interoperability of genomic data and EHR, freeing its use for clinical care and research. It will also allow stakeholders to analyze real-world data from large cohorts of patients. Access to data on patients with diverse demographics, allows informed treatment decisions between clinicians and patients, new research, and the development of clinical decision support tools for clinical use, and to meet the needs of other

Potential focus of initial use case

Members who are part of the Use Case will determine the initial types of genomic reporting to focus on. Examples of tests might include ACMG 59 Actionable Disorders Panel, Oncology focused genomic testing, Whole Genome or Exome Sequencing test. Use Case Members will then develop scenario-based profiles/import specifications leveraging the FHIR Genomics IG to develop communication of structured genomic data from a laboratory to a receiving organizations EHR or genomic data repository. Use Case members will include both creators and recipients of FHIR Genomics data in order to ensure that specifications are straightforward to construct, and that the data can be correctly parsed and processed on the receiving end.