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Implementation of Gene Expression Profiles in the HL7 FHIR Standard

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Abstract. Gene expression profiles can capture significant molecular differences paving the way toward precision medicine. However, clinical standards like FHIR only provide encoding of molecular sequence variations, even so, expression patterns are equally important for decision making. Here we provide an exemplary implementation of gene expression profiles of a microarray analysis using an adaption of the FHIR Genomics extension. Our results demonstrate how FHIR resources can be facilitated in bioinformatics-based decision support systems or used for the aggregation of molecular genetics data in multi-center clinical trials.

Keywords. FHIR, interoperability, omics, gene expression

1. Introduction

Over the years, interoperability gained more importance in clinical settings with standards like Fast Healthcare Interoperability Resources (FHIR) [1]. However, the genomic profiles in the FHIR Genomics extension only cover variations in the molecular sequence while expression patterns are neglected. Nevertheless, insights from expression patterns are important for decision support and translational medical research. Here we present a feasible FHIR implementation for gene expression profiles.

2. Methods

The central element within FHIR to capture real-world concepts in healthcare systems is the *Patient* resource, which is why all subsequent patient-specific results and resources were referred to this base. For the preservation of the anonymity of participants, we used artificially generated data to create *Patient* resources using Synthea™ [2]. The medical condition was captured by the *Condition* and samples by the *Specimen* resource to allow multiple samples for a single patient. Referenced genes were included as *MolecularSequence* resources, which avoids redundancy in common genomic profiles and simplifies retrieval of expression values for a single gene across multiple samples. The expression values are treated as single measurements of *Observation-geneticsGene* resources referring to the corresponding gene. An overview is shown in figure 1.

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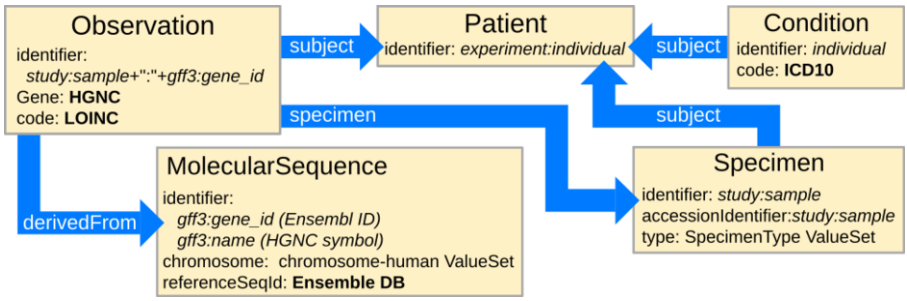

Figure 1. Overview of the extended FHIR resources and references for gene expression profiles.

3. Results

For demonstration, we utilized a HAPI FHIR server [3] and an mRNA microarray [4], which examines a dose-limiting side effect of chemotherapy in patients with acute myeloid leukemia (EBI Expression Atlas: E-GEOD-10746). The data set translated to 252,684 resources stored on our FHIR server. Our implemented FHIR solution is available on GitHub: https://github.com/frankkramer-lab/gene-expression-on-fhir.

4. Discussion

Through our contribution to the FHIR Genomics extension, we were able to include genomic profiling data allowing enhanced clinical interoperability of genomic data. We are planning further evaluation by application in a decision support system as well as consolidation of the outcome of expression analyses as *DiagnosticReport* resources.

5. Conclusions

Our results demonstrate how FHIR resources can be facilitated for the clinical exchange of expression profiles. The further extension of FHIR allows for establishing a currently missing standard for molecular genetics data in clinical settings.

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