HITSAC Clinical Genomics Use Case & Pilot Project: Pharmacogenomics

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www.vita.virginia.gov
Abstract

• Defines the process for integrating results from pharmacogenomic (PGX) testing across disparate electronic health record (EHR) systems

• Developed by the Virginia Information Technologies Agency (VITA) at the direction of HITSAC and the HITSAC Genomics Working Group (2014)

• Partners include the Virginia Department of Health (VDH), Inova Health System, Inova Translational Medicine Institute (ITMI), the Sequoia Project/eHealth Exchange, and Virginia Commonwealth University (VCH) Health System
Background

- PGX explores the role of genetics in drug response, focusing on the influence of genetic variation on drug response in patients by correlating gene expression or single-nucleotide polymorphisms with drug absorption, distribution, metabolism and elimination and drug receptor target effects.

- PGX concentrates on single drug-gene interactions and encompasses a more genome-wide association approach, incorporating genomics and proteomics while exploring the effects of multiple genes on drug response.
Background

• Results from PGX testing require a designated location, standardized structure and nomenclature, and dedicated persistence within the EHR.

• Unlike other laboratory tests, results from PGX testing do not change.

• PGX test results therefore must remain with the patient and be clearly discoverable by health care providers to inform appropriate pharmacological treatment.
PGX Test Result Requirements

- A clinical report with the results described in narrative form
  - Actions and recommendations signed off by the certified clinical and or molecular geneticists
  - Understandable information for both clinician and patient
PGX Test Result Requirements

- A discrete result of the allele variation for the specific loci being tested
  - Allows for interoperability and data sharing based on a standard set understandable across the genetic community
  - Enables alerts – both active and passive – to be generated based on the results
  - Provides for statistical and outcomes-based reporting from the results
  - Supports future reporting and alerts based on discovery of validated drug/gene interactions
• A discrete, more descriptive result of the test in the context of the test order
  – Provides for statistical and outcomes-based reporting for non-genetic purposes
  – Gives clear indication result interpretation in the context of patient point of care
PGX Pilot Project

- Developed by HITSAC to define the process, and associated health IT standards, for integrating results from PGX across disparate EHR systems.
- Leverages the workflow, testing protocols, and reporting capability currently being implemented by Inova/ITMI as the Plavix Genotype Test.
- Goal will be to transmit the results of Inova/ITMI PGX testing into Inova’s EPIC HER system then to VCU Health System’s Cerner EHR system through eHealth Exchange.
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<tr>
<th>ID</th>
<th>TITLE</th>
<th>NOTES</th>
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<tr>
<td>EDS-R-163 (COV)</td>
<td>HL7/Logical Observation Identifiers Names and Codes (LOINC)</td>
<td>LOINC #81247-9: HL7 Genetic Variant Reporting Panel (See attached panel hierarchy)</td>
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<tr>
<td>EDS-R-47 (COV)</td>
<td>National Center for Biotechnology Information (NCBI) Genetic Reference Sequences</td>
<td>NCBI RefSeqGene</td>
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<td>CAQH CORE X12</td>
<td>Council for Affordable Quality Health (CAQH) Care Committee on Operating Rules for Information Exchange (Phase I and Phase II)</td>
<td>CAQH CORE X12 Document Submission Service Interface Specification v. 1, ANSI X12, required for eHealth Exchange testing, certification, and onboarding</td>
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<td>NHIN WSR/WSI</td>
<td>Nationwide Health Information Network (NHIN) Web Services Registry Web Service Interface Specification, Version 3.1</td>
<td>NHIN WSR/WSI Specification required for eHealth Exchange testing, certification, and onboarding</td>
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<td>NHIN X12 esMD</td>
<td>Nationwide Health Information Network (NHIN) Electronic Submission of Medical Documentation (esMD) X12 Profile, Version 1</td>
<td>ANSI X12 esMD required for eHealth Exchange testing, certification, and onboarding</td>
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<td>HL7 V2IG CG LOINCGENVAR R2-2013</td>
<td>HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 2</td>
<td>Ref. HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1</td>
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<td>HL7 ORU^R01</td>
<td>HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1</td>
<td>Defines necessary specifications for clinical laboratory results reporting to EHRs for use in the U.S. Realm</td>
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<td>Project Task</td>
<td>Description</td>
<td>Due Date / HITSAC Status Report</td>
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<td><strong>Task 1: Stakeholder Engagement</strong></td>
<td>Coordination among stakeholders to level-set on use case and project objectives; trust framework execution (if required); documentation of stakeholder systems and exchange specifications</td>
<td>01/31/2017 / 01/19/2017</td>
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<td><strong>Task 2: Requirements &amp; Specifications Analysis</strong></td>
<td>Analysis of use case requirements, system specifications, performance and service specifications, and security/privacy provisions; documentation of requirements and specifications to guide project onboarding, testing, certification, and implementation (<a href="http://sequoiaproject.org/resources/exchange-specifications/">http://sequoiaproject.org/resources/exchange-specifications/</a>)</td>
<td>03/31/2017 / 03/16/2017</td>
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<td><strong>Task 3: Onboarding, Testing, &amp; Certification</strong></td>
<td>Completion of required onboarding, testing, and certification of stakeholder systems onto the eHealth Exchange (<a href="http://sequoiaproject.org/ehealth-exchange/onboarding/">http://sequoiaproject.org/ehealth-exchange/onboarding/</a>)</td>
<td>05/30/2017 / 05/18/2017</td>
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<td><strong>Task 4: Implementation and Production Exchange</strong></td>
<td>Implementation of exchange in a production environment; ongoing monitoring and exchange refinement during project’s period of performance</td>
<td>07/31/2017 / 07/20/2017</td>
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<td><strong>Task 5: Process, Data &amp; Transaction Workflow Review</strong></td>
<td>Comprehensive review of business processes, data flows, and transaction workflows in production environment; documentation of review to exchange with HL7 and other external stakeholders</td>
<td>09/30/2017 / 09/21/2017</td>
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Requested HITSAC Action

- Direct HITSAC staff to work with Virginia Department of Health and other stakeholders to implement the proposed work plan and schedule
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