Virginia Information Technologies Agency

HITSAC Clinical Genomics Use Case & Pilot Project: Pharmacogenomics

Joseph W. Grubbs, Ph.D.
Staff Analyst

Presentation to the
Health IT Standards Advisory Committee
November 10, 2016

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

PGX Test Result Requirements

- 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

PGX Pilot Project – Health IT Standards

ID	TITLE	NOTES
EDS-R-163 (COV)	HL7/Logical Observation Identifiers Names and Codes (LOINC)	LOINC #81247-9: HL7 Genetic Variant Reporting Panel (See attached panel hierarchy)
EDS-R-47 (COV)	National Center for Biotechnology Information (NCBI) Genetic Reference Sequences	NCBI RefSeqGene
CAQH CORE X12	Council for Affordable Quality Health (CAQH) Care Committee on Operating Rules for Information Exchange (Phase I and Phase II)	CAQH CORE X12 Document Submission Service Interface Specification v. 1, ANSI X12, required for eHealth Exchange testing, certification, and onboarding
NHIN WSR/WSI	Nationwide Health Information Network (NHIN) Web Services Registry Web Service Interface Specification, Version 3.1	NHIN WSR/WSI Specification required for eHealth Exchange testing, certification, and onboarding
NHIN X12 esMD	Nationwide Health Information Network (NHIN) Electronic Submission of Medical Documentation (esMD) X12 Profile, Version 1	ANSI X12 esMD required for eHealth Exchange testing, certification, and onboarding
HL7 V2IG CG LOINCGENVAR R2-2013	HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 2	Ref. HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1
HL7 ORU^R01	HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1	Defines necessary specifications for clinical laboratory results reporting to EHRs for use in the U.S. Realm

PGX Pilot Project – Proposed Work Plan

Project Task	Description	Due Date / *HITSAC Status Report*
Task 1: Stakeholder Engagement	Coordination among stakeholders to level-set on use case and project objectives; trust framework execution (if required); documentation of stakeholder systems and exchange specifications	01/31/2017 / *01/19/2017*
Task 2: Requirements & Specifications Analysis	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 (http://sequoiaproject.org/resources/exchange-specifications/)	03/31/2017 / *03/16/2017*
Task 3: Onboarding, Testing, & Certification	Completion of required onboarding, testing, and certification of stakeholder systems onto the eHealth Exchange (http://sequoiaproject.org/ehealth-exchange/onboarding/)	05/30/2017 / *05/18/2017*
Task 4: Implementation and Production Exchange	Implementation of exchange in a production environment; ongoing monitoring and exchange refinement during project's period of performance	07/31/2017 / *07/20/2017*
Task 5: Process, Data & Transaction Workflow Review	Comprehensive review of business processes, data flows, and transaction workflows in production environment; documentation of review to exchange with HL7 and other external stakeholders	09/30/2017 / *09/21/2017*

Requested HITSAC Action

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

For More Information

Joseph W. Grubbs, Ph.D.
HITSAC Staff Analyst
Virginia Information Technologies Agency
Phone: (804) 467-7729
Email: *Joseph.Grubbs@vita.virginia.gov*