

**Progress on Health IT
Standards for Clinical
Genomics**

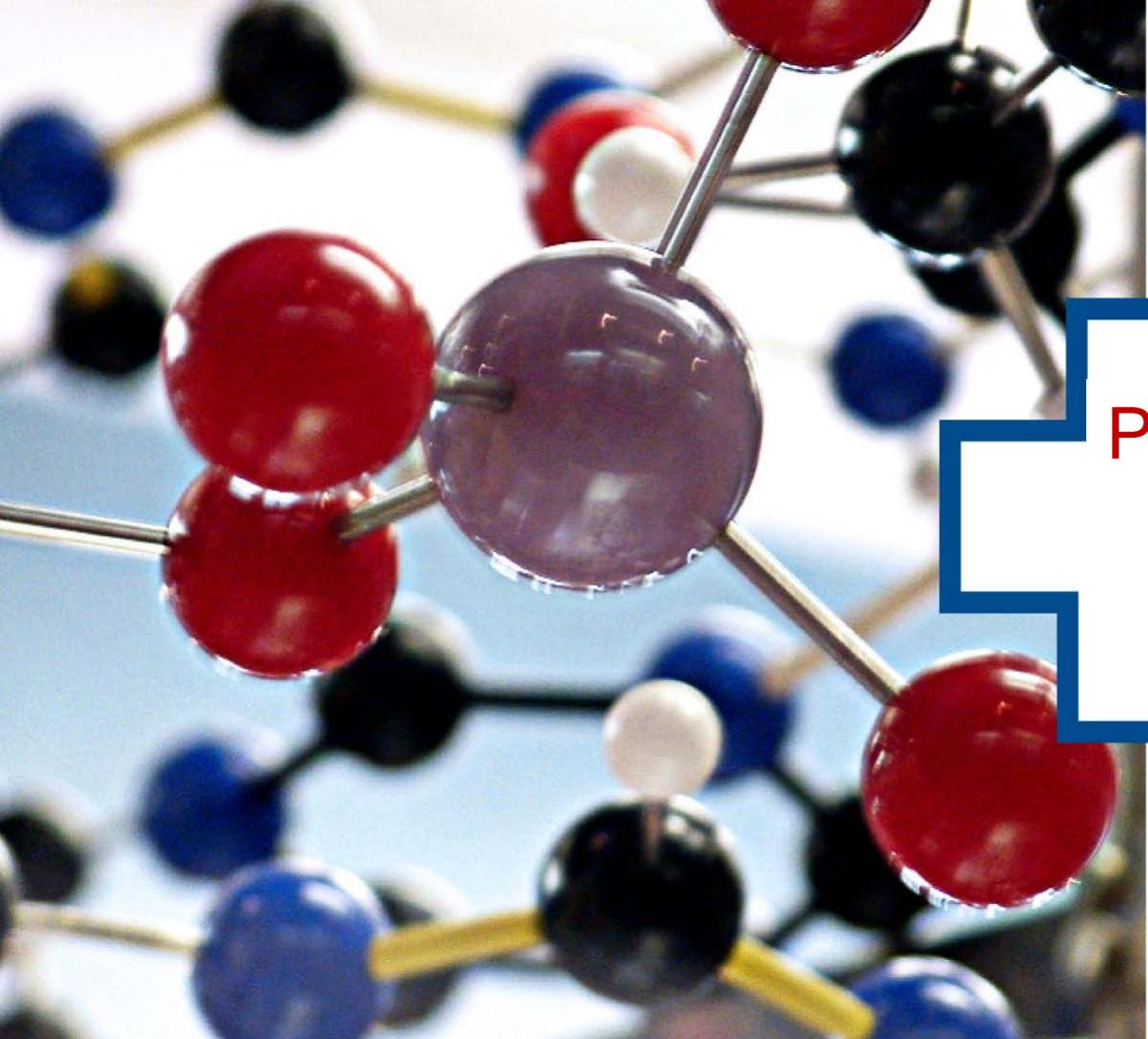
**Health Information Technology
Standards
Advisory Committee (HITSAC)**

*Aaron Black
Director of Informatics
Inova Translational Medicine Institute
September 29, 2016*

- Summary of Previous Presentation on May 12, 2016
- Updates Since
- Process Recommendations
- Action items
- Questions and Comments

- Genetics and Genomics data standards still in process for clinical results
 - Many sets of standards
 - Pharmacogenomics standards are simpler can closer to final than standards for cancer genomics
- HL7 and LOINC are working together on standards
 - Next update in Fall 2016 – HL7 and LOINC are working together well and getting close to shared standards for genomics – still very complicated. HL7 gives us standards for interfaces and LOINC gives us standards for coding – their work complements each the other.
 - Inova has begun to adopt for Pharmacogenomics testing
- Virginia can be leader for application of standards – with HITSAC and its focus on Genomics
- Recommendation
 - Create process for genetic \ genomic testing standardization - working with ITAC and CIO of Commonwealth, State Laboratories, Virginia Department of Health, health care systems in Virginia
 - Create multi-disciplinary team to create and manage process
 - People from State Labs, VDH, laboratorians, bioinformaticists, clinicians will be needed on the multi-disciplinary committee
 - Use real-world implementations as companion process for better evaluation

- Inova team met at the National Library of Medicine in June, 2016, with LOINC founder (Clem McDonald, MD), HL7 Genomics data standards committee members (Mollie Ullman-Cullere, PhD, and national software vendor (Don Rule, CEO of Translational Software) for detailed discussion on proposed LOINC and HL7 updates to be approved in Fall of 2016
- Standards are still being debated, though more thorough than previous standards
 - Repeating variants and collections of variants are challenging to identify in codes; we need to add appropriate dosage of medications with results of pharmacogenomic tests to drive alerts for physicians prescribing medications.
- Key discussion items included:
 - Structure of HL7 messaging for single and complex gene results.
 - LOINC will update coding of various coded elements
 - Discussed how data standards need to accommodate enough information in order to:
 - Enable clinical decision support within EHR's
 - Enable electronic interaction with internal and external knowledgebase systems to
 - keep results relevant and actionable as knowledge accumulates
 - become more detailed
 - How to interact and give feedback to improve the standards.
 - How to distinguish germ line from somatic line genes and changes from reference genome



Proposed Process for Genetic \ Genomic Standards

- Current Implementation Roadmap Recommendations (used by several pilot organizations) – for health systems and vendors of EMRs
 1. Incorporate design in databases
 2. Implement design standard in laboratory reports and/or data files
 3. Validate utility of information model through active use in business
 4. Iterate on information model incorporating lessons learned
 5. Formally develop HL7 interfaces for fully codified/qualified data when business is ready

- **Proposed process**

(1) Organize a multi-disciplinary and cross-functional team to build methodology on data specifications for Genetic and Genomic tests – Virginia can lead the USA in this effort!

– Who should be involved?

- Health Systems \ Academic Medical Centers \ State Labs (2-3 distinct institutions)
 - Criteria
 - » Have or will offer genetic \ genomic tests
 - » Lab \ Genetic Experts (humans, microbes, animals)
 - » Dedicate Clinical, Genomic and EHR expertise
 - » Informatics experts
- Virginia Government
- External informatics experts (HL7 and LOINC)
- Software vendor(s) – such as EMR vendors and Translational Software

(2) Goal for multi-disciplinary and cross-functional team

- Create standards recommendations to Virginia for at least 2 genetic tests
- Select:
 - One simple test
 - One slightly more complex
- An example would be Pharmacogenomics (PGx)
 - Simple → Individual Gene \ Drug test (ie Plavix or Warfarin)
 - More complex → Set of PGx genes and alleles (ie MediMap)
 - Cancer genomics are too complicated now, let's start with pharmacogenomic testing
- About one-third of persons will have a significant unexpected “abnormality” in their results of pharmacogenomic tests – meaning the metabolize common medications in unexpected ways that their physicians should know.

(3) For those tests, the team would present specification to HITSAC recommendations on, but not limited to the following:

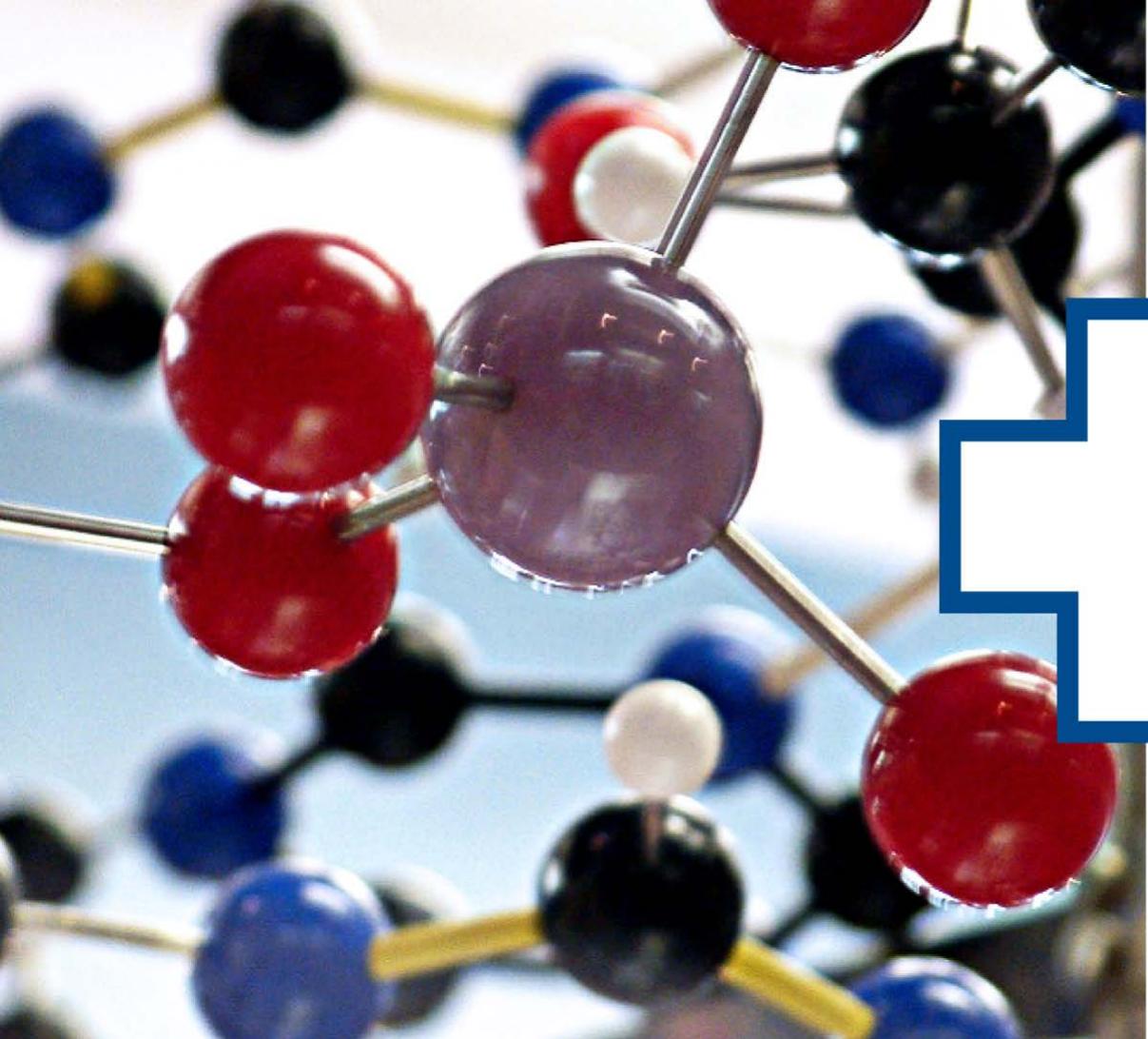
- Coding and messaging standards (LOINC, HGVS, COSMIC..)
 - HGVS: Human Genome Variant Standards; COSMIC: genetic standards for cancer
- Data exchange standards between like institutions (LOINC & HL7)
- Reporting standards
 - Return of results to EHR (Formats and exchange \ LOINC, HL7)
 - Impact of tests – we need some team to perform economic analysis on the value of these tests and standards
 - » Patient care improvement (short and medium term)
 - » Return on investment – to patients, sources of tests and insurers
 - » Overall efficiencies gained by avoiding adverse drug events and ineffectual medications in specific persons.

(4) Present roadmap for future tests to evaluate standards for more complex and diverse tests.

- To support clinical use cases such as these:
 - Newborn screening – shall we do genetic testing for all newborns, across Virginia?
 - Prenatal screening
 - Cancer treatment \ Clinical trials \ Cancer Registry
 - Rare disease
 - Public Health Reporting \ Virginia Health Information Exchange
- Type of tests \ technologies
 - Panels – Next Generation Sequencing (NGS) \ Array and others
 - Whole Exome (from reference genome giving mapping of nucleotides to genes)
 - Whole Genome Sequencing – plummeting in cost
 - Proteomics \ Cytogenetic testing

- Complex standards need experts from multiple disciplines
 - This is too large a project for one organization – if we can work together we can establish standards for pharmacogenomic testing while we wait for LOINC and HL7 to finalize standards for cancer genomics.
- Agility will be key, as tests and national standards are adopted and change
 - Standards from LOINC & HL7 will change – and we will need to upgrade standards as they mature
- Start with simple tests. The standards are better defined for the simple tests
 - Pharmacogenomics – single genes and multi-gene panels
- Application of these standards in one or more institutions will help filter out real world issues and create momentum.
 - Inova, UVA, Sentara, VCU, Carilion, Bon Secours, HCA – to name a few – can pilot these standards
- This volunteer effort is needed!

- Discuss
 - Participants
 - Genetic Tests to be evaluated
 - Communication plans
 - Next Steps



Q&A

Comments

