

Health IT Standards Committee

A Public Advisory Body on Health Information Technology to the National Coordinator for Health IT



Precision Medicine Task Force

Preliminary Recommendations to Health IT Standards Committee

September 22, 2015

Leslie Kelly Hall, Co-Chair

Jon White, Co-Chair



- **Precision Medicine Initiative**
- **Task Force Charge, Work Plan, Membership**
- **Review and Finalize Recommendations**
- **Next Steps**

Mission Statement

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized treatments.





- Identify opportunities for innovative collaboration around pilots and testing of standards that support health IT interoperability for precision medicine
- Recommend existing standards that are currently ready to support PMI
- Identify emerging standards and reference implementations that may require further pilot testing in order to support PMI
- Identify gaps in available data standards related to PMI

Precision Medicine Task Force Workplan



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	Meetings	Task
✓	July 17, 2015 11:00 am - 1:30 pm ET Kick-off	<ul style="list-style-type: none"> Review charge, work plan Overview of the Precision Medicine Initiative - White House Office of Science & Technology Policy & National Institutes of Health
✓	Wednesday Jul 29, 2015 1:30 - 3:00 pm ET	<ul style="list-style-type: none"> Presentations from experts - 23andMe; NIH Precision Medicine Workshop; Institute of Medicine Genomics Roundtable
✓	Wednesday Aug 5, 2015 12:00 - 1:30 pm ET	<ul style="list-style-type: none"> Presentations from experts - Intel Corporation; Intermountain Healthcare; National Library of Medicine
✓	Wednesday Aug 19, 2015 12:00 - 1:30 pm ET	<ul style="list-style-type: none"> Presentations from experts - Duke Me Tree Project; eMerge Network; New York Genome Center; Sage Bionetworks
✓	Monday Aug 31, 2015 12:00 - 1:30 pm ET	<ul style="list-style-type: none"> Develop Preliminary Task Force Recommendations – Broad Institute
✓	Thursday Sept 10, 2015 1:30 - 3:00 pm ET	<ul style="list-style-type: none"> Revise Task Force Recommendations
✓	Friday Sept 18, 2015 Time TBA	<ul style="list-style-type: none"> NIH ACD Update HL7 Genomics Workgroup Finalize Recommendations to HITSC
→	September 22, 2015 – HITSC Meeting 10:00 am – 12:30 pm ET	<ul style="list-style-type: none"> Present final recommendations

Precision Medicine Task Force Membership



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Member	Organization
Co-Chairs	
Leslie Kelly Hall	Healthwise
Jon White	ONC / Agency for Healthcare Research and Quality (AHRQ)
Members	
Mary Barton	National Committee for Quality Assurance (NCQA)
Lisa Gallagher	Healthcare Information and Management Systems Society (HIMSS)
David McCallie, Jr.	Cerner Corporation
Andrey Ostrovsky	Care at Hand
Eric Rose	Intelligent Medical Objects
Andrew Wiesenthal	Deloitte Consulting, LLP
Federal Ex Officio	
James Breeling	Veterans Health Administration (VHA)
Josh Denny	National Institutes of Health (NIH)
Christina Heide	HHS Office for Civil Rights (OCR)
Betsy Humphreys	National Library of Medicine (NLM)
Mitra Rocca	Food and Drug Administration (FDA)
Invited Guests	
Mina Hsiang	United States Digital Service (USDS) / Office of Management and Budget (OMB)
Claudia Williams	White House Office of Science & Technology Policy
ONC Staff	
Maya Uppaluru	ONC – Federal Staff Lead
Debbie Bucci	ONC - Technical Advisor



Draft Recommendations



- **Background** - EHR likely to capture more phenotypic data from MDs and patient
 - Phenotypic data are collected already such as problems, medications, allergies, etc.
 - Core problem: Don't have a standard data model for EMR and categorical standard responses for many basic types of information
- **Standards and Recommendations** were placed into four categories:
 1. **Readily Applicable Standards for PMI (Green)** – can be put to use to support the cohorts
 2. **Promising Standards for PMI (Yellow)** – may require additional effort to bring to use
 3. **Standards Gaps for PMI (Red)** – areas where considerable work is needed
 4. **Accelerators (Blue)** – opportunities to advance / improve standards
- **Recommended Actions to Advance need to be assigned** to each standard, emerging standard, recommendation, etc.
 - Short-term versus long-term priorities

Readily Applicable Standards for PMI



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Recommendation	Actions to Advance
Precision Medicine efforts should align to standards currently referenced in the 2015 Interoperability Standards Advisory where they are included in current regulation, including EHR Incentive Program and Health IT Certification Rules (e.g. race & ethnicity, family health history/clinical genomics, gender identity, sex and sexual orientation, patient preference/consent)	C
Use standards to capture and represent family health history such as SNOMED CT and the HL7 family health history and pedigree model for familial relationships, in order to express as a pre-coordinated or post-coordinated code	B
Leverage HL7 Clinical Genomics WG suite of standards (CDA, FHIR, v2, domain analysis model)	B
Key: Actions to Advance A – Form Task Force to advance for PMI B - Apply accelerators (e.g., S&I Initiative, pilot project, policy guidance) to existing standards by ONC C - Follow existing standards process	

Promising Standards for PMI



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Recommendation	Actions to Advance
<p>FHIR could be included as an emerging standard⁴, especially for transport of data. Argonaut may provide opportunities to advance. Sample uses of FHIR: authorization; genetics, family health history, build on current work on SMART on FHIR Genomics¹⁾</p>	<p>B</p>
<p>Open ID Connect, OAuth and UMA should be considered for single sign on; further piloting and testing should be considered</p>	<p>B</p>
<p>Computable Patient Consent: standards exist but lack adequate implementation guidance³ (Lower priority will be handled separately from the standards process)</p>	<p>C</p>
<p>Include more complete authorization standards (e.g., IHE XUA, IUA, etc.); ensure authorization standards are compatible across disparate networks⁵</p>	<p>C</p>
<p>IOM Genomics Roundtable, GA4GH work</p>	<p>B</p>

Key: Actions to Advance

A – Form Task Force to advance for PMI

B - Apply accelerators (e.g., S&I Initiative, pilot project, policy guidance) to existing standards by ONC

C - Follow existing standards process

1) Precision Medicine Task Force: <http://healthit.gov/FACAS/calendar/2015/08/19/precision-medicine-task-force>

2) Precision Medicine Task Force: <http://healthit.gov/FACAS/calendar/2015/08/31/precision-medicine-task-force>

3) Interoperability Standards Advisory Task Force - http://healthit.gov/FACAS/sites/faca/files/HITSC_ISATF_Recommendation_Slides_2015-08-26.pdf page 17

4) Ibid page 19

5) Ibid Page 19

Standards Gaps for PMI



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Recommendation	Actions to Advance
ONC should convene a stakeholder group to address computable patient consent; there exist standards but without clear implementation guidance ¹	B
Race and Ethnicity: OMB Standard may be suitable for some purposes but inadequate for precision medicine and directing therapy or clinical decisions ²	A
ONC should work with stakeholders to define what is the minimum data set and/or means required to make precision medicine data useful in an EHR and in a clinical setting ³	A
Microbiome data standards ⁴	C
Capture of sexual orientation and gender identity remain challenging, ONC should consider recent efforts of the Fenway Institute in this area	B

Key: Actions to Advance

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B - Apply accelerators (e.g., S&I Initiative, pilot project, policy guidance) to existing standards by ONC

C - Follow existing standards process

1) Interoperability Standards Advisory Task Force – Page 17 http://healthit.gov/FACAS/sites/faca/files/HITSC_ISATF_Recommendation_Slides_2015-08-26.pdf

2) Ibid. page 15

3) Precision Medicine Task Force: <http://healthit.gov/FACAS/calendar/2015/08/05/precision-medicine-task-force>

4) Precision Medicine Task Force: <http://healthit.gov/FACAS/calendar/2015/08/31/precision-medicine-task-force>



Recommendation	Actions to Advance
2016 PMI S&I: Additional ONC investment in pilots of FHIR for PMI research/individual data donation use case	B
Incorporation of HPO in the UMLS Metathesaurus and connections between HPO and SNOMED CT ²	C
OMIM: Codes for phenotypes, genotypes and links between the two	C
dpSNP and ClinVar: Opportunity to develop a service that would consumer data from these sources and synthesize so it's digestible for a clinical information system	C

Key: Actions to Advance

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1) Interoperability Standards Advisory Task Force – Page 19 http://healthit.gov/FACAS/sites/faca/files/HITSC_ISATF_Recommendation_Slides_2015-08-26.pdf

2) <http://ebooks.iospress.nl/publication/40319> , <http://mor.nlm.nih.gov/pubs/pdf/2015-phenoday-fd.pdf>



Next Steps



Appendix



Focus

- The exchange of genomic and phenomic data among:
 - Patients/Participants
 - EHRs
 - Researchers
 - Testing labs (for both research and clinical care)

Questions for Presenters

- What is the key problem or set of problems your organization is attempting to solve?
- What is the minimum interoperable data set of genome and phenome data for these data exchanges?
- Are there standards that can support this movement today?
- What gaps are there and what is needed in the future?