September 25, 2015

Karen DeSalvo, MD
National Coordinator for Health Information Technology
Department of Health and Human Services
200 Independence Avenue, SW
Washington, DC 20201

Dear Dr. DeSalvo,

In response to the recommendations from the Precision Medicine Task Force, the Health Information Technology Standards Committee (HITSC) was asked to provide your office with recommendations around the standards to support the President's Precision Medicine Initiative. This transmittal offers these recommendations, which are informed by the deliberations among the Task Force subject matter experts, and presentations from relevant stakeholders.

Background:

In the State of the Union in January 2015, President Obama announced the launch of the Precision Medicine Initiative (PMI), with the mission 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. PMI is an inter-agency effort, including important work from the National Institutes of Health, the Food and Drug Administration, and the Office of the National Coordinator for Health IT. ONC's role to support PMI is to recommend policies and standards to support privacy and security of participant data, as well as standards that support a participant-driven approach to data contribution, and identify opportunities for innovative collaboration around pilots and testing of standards that support health IT interoperability for research.

In furtherance of this mandate, the Precision Medicine Task Force came together in July 2015 with the following charge:

- 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

The Task Force presented its recommendations to the HITSC on September 22, 2015.[1] In that meeting, the HITSC approved the below recommendations of the Task Force.

The recommendations, presented herein, respond to the Task Force Charge.

Recommendations:

Standards and Recommendations were placed into four categories:

- Readily Applicable Standards for PMI (Green): Standards that have been tested and implemented in the health IT market that can be put to use to support the NIH PMI Cohort and precision medicine.
- Promising Standards for PMI (Yellow): Standards that may require additional effort, including further pilots and testing, to support precision medicine.
- Standards Gaps for PMI (Red): Areas where considerable additional standards development work is needed.
- Accelerators (Blue): The Task Force also identified opportunities to advance and improve the standards that fell in the previous categories.

Actions to Advance were also assigned to each recommendation:

A – ONC should form an additional Task Force to advance this recommendation for PMI

B – ONC should apply various tools (e.g., S&I Initiative, pilot project, policy guidance) to advance these existing or emerging standards

C – ONC should follow and monitor existing standards development processes

Readily Applicable Standards for Precision Medicine

- 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 (C)
- Use standards to capture and represent family health history such as SNOMED CT and the HL7 Version 3 Implementation Guide: Family History/Pedigree for familial relationships, in order to express as a pre-coordinated or post-coordinated code (B)
- Leverage HL7 DIGITizE Actions Collaborative draft LOINC specification for pharmacogenomics by supporting ongoing IOM Genomic Roundtable efforts (B)

[1] http://www.healthit.gov/facas/FACAS/calendar/2015/03/18/hit-standards-committee

Promising Standards for Precision Medicine

- Support HL7 Clinical Genomics WG standards development, including CDA R2 Clinical Genetics Reporting, Clinical Genomics Pedigree Model, HL7 Genetic Testing Results Message (V2), and Clinical Sequencing Domain Analysis Model (DAM) (B)
- Open ID Connect, OAuth and UMA should be considered for authorization and authentication; further piloting and testing should be considered (B)
- Include more complete authorization standards (e.g., IHE XUA, IUA, etc.); ensure authorization standards are compatible across disparate networks (C)
- Support Global Alliance for Genomics and Health (GA4GH) work to address computable consent in research context (C)

Standards Gaps for Precision Medicine

- ONC should convene a stakeholder group to address granular, dynamic computable consent. There are existing standards in this space, but without clear implementation guidance, and alignment between HIPAA and Common Rule should be addressed (A)
- Race and Ethnicity: OMB Standard may be suitable for some purposes but inadequate for precision medicine and directing therapy or clinical decisions (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 (A)
- Microbiome, exposome data standards (C)
- Capture of sexual orientation and gender identity remain challenging; ONC should consider how to advance recent efforts of the Fenway Institute in this area (B)

Accelerators

- 2016 Precision Medicine Initiative pilots: ONC should invest in pilots of FHIR to support individual data donation use case, which is centrally important to the Precision Medicine Initiative and NIH Cohort (B)
- Support incorporation of Human Phenotype Ontology (HPO) in the Unified Medical Language System (UMLS) Metathesaurus and connections between HPO and SNOMED CT (C)
- Support ongoing work Online Mendelian Inheritance in Man (OMIM): Codes for phenotypes, genotypes and links between the two (C)
- Support dbSNP and ClinVar: Opportunity to develop a service that would get consumer data from these sources and synthesize so it's digestible for a clinical information system (C)

We appreciate the opportunity to provide these recommendations and look forward to discussing next steps.

Sincerely yours,

/s/

P. Jon White

Chair, Health IT Standards Committee

/s/

John D. Halamka

Vice Chair, Health IT Standards Committee