Precision Medicine Initiative

Next Steps

December 10, 2015

Precision Medicine Initiative

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.

ONC Role in Precision Medicine

- Recommend policies and standards to support privacy and security of participant data
- Identify standards that support a participant-driven approach to data contribution
- Coordinate with others to identify opportunities for innovative collaboration around pilots and testing of standards that support health IT interoperability for research

Precision Medicine Task Force
Final Recommendations

PMI Task Force Recommendations

- **Presented recommendations to HITSC Sept 22 and to Joint Committee on Oct 6**
- **Background** - EHR likely to capture more phenotypic data from MDs and patient
  - Phenotypic data are collected already such as problems, medications, allergies
  - 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 were assigned** to each standard, emerging standard, recommendation

Readily Applicable Standards for PMI

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	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 DIGITizE Actions Collaborative draft LOINC specification for pharmacogenomics by supporting ongoing IOM Genomic Roundtable efforts	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

1) Transport & Security Standards Workgroup: http://healthit.gov/facas/calendar/2014/10/08/standards-transport-security-standards-workgroup

Promising Standards for PMI

Recommendation	Actions to Advance
Support HL7 Clinical Genomics WG standards development (CDA, v2, domain analysis model, SMART on FHIR Genomics)	B
Open ID Connect, OAuth and UMA should be considered for authentication and authorization; 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[5]	C
Support Global Alliance for Genomics and Health (GA4GH) work to address computable consent in research context	B

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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) Interoperablity 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

Recommendation	Actions to Advance
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.[1]	A
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, exposome, etc 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) Interoperablity 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

Accelerators

Recommendation	Actions to Advance
2016 PMI use case/pilots: Additional ONC investment in pilots of FHIR for PMI research/individual data donation use case	B
Support incorporation of HPO in the UMLS Metathesaurus and connections between HPO and SNOMED CT[2]	C
Support ongoing OMIM work: Codes for phenotypes, genotypes and links between the two	C
Support 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

- How can ONC support the use of emerging standards (FHIR, OAuth 2) to support individual data donation to PMI?
- What is the best way to execute on the Task Force's recommendation that ONC convene stakeholders to address dynamic computable consent?
- What existing work can ONC support to improve race and ethnicity standards and capture of sexual orientation and gender identity data, so that they are adequate for precision medicine and directing therapy or clinical decisions?
- How should ONC execute on the recommendation to define the minimum data set and/or means required to make precision medicine data useful in a clinical setting?