

NGS & Standards

- Concerns over NGS standards
 - Includes bioinformatics
- IgDAWG is investigating these
 - <http://igdawg.org>
- Aim to develop standards and pilot these in the NGS workshop component
 - MIRING

FDA NGS Standards Workshop: Recap

- NGS standards (FDA Genomics Working Group, HL7, NCBI)
- Big data administration & computational infrastructure
 - (FDA/CBER/HIVE, NCI, NY Genome Center)
- Database development
 - (NCBI, Stanford, NCBI/Virus Genome Group)
- Biologics product evaluation
 - (FDA/CBER, UCSF, Sanofi Pasteur, Merck)
- Clinical biomarkers and personalized medicine
 - (VCUMC, Celera/Quest, MSKCC, UCSF)
- NGS sequencing devices and clinical applications
 - Panel - PacBio, Ion Torrent, Illumina, Complete Genomics
- Food safety and pathogen detection (FDA, NCBI, CDC)

- Nothing new from FDA in terms of regulations or even guidelines
 - They are gathering info from community
 - Some people complaining; others working on solutions



DEPARTMENT OF HEALTH & HUMAN SERVICES

FOOD AND DRUG ADMINISTRATION

Office of Director
Center for Biological Evaluation and Research

**Next-generation sequencing (NGS)
technology, data formats standardization
and promotion of interoperability protocols**



Date and Time: September 24-25, 2014 from 8:30 a.m. to 5:00 p.m.

Data Standards Hackathon (DaSH) Recap

- MIRING revised substantially: reduced from 10 to 8 items
- HML:
 - released versions 0.9.4, 0.9.5, 0.9.6
 - incorporated MIRING features
 - HML messages created/processed by many
 - Implementation issues (performance, security)
- Addressed questions about GL String, GL Service, OIDs, GTR,
- Finalized specification for a Multiple-Allele Code Creation Service

Next Steps

- Address “issues” identified
- Publish HML, MIRING and meeting report
- Special issue of Human Immunology on NGS coming early next year
- Use NGS component of the workshop as a pilot for the standards

Extract sequence of interpretable regions from filtered assembly consensus sequences

Verify assembly consensus sequences align within targeted region

How do we evaluate alignment of paired reads against genome reference assembly?

Interpret “all exons” extracted from assembly consensus sequences

Are paired reads adaptor trimmed? quality trimmed?

Interpret whole gene assembly consensus sequences

Calculate coverage per targeted region by assembly consensus sequences

Filter alignment of assembly consensus sequences against genome reference assembly for interpretable regions (BED or GFF format?)

Align assembly consensus sequences against genome reference assembly and alternative assemblies (MHC, KIR)

What is clinically interpretable?