IT/Bioinformatics & CDS

- Did not consider anything prior to creation of .vcf file.
- Discussed existing and feasible standards that would be desirable to, as a group, move toward.
- Avoid mapping different standards to a created standard unless absolutely necessary.

IT/Bioinformatics & CDS – Prioritized Actions

1. What is connection between genotype and phenotype? (0/0)
2. Define key elements that should be stored in EHR. (11/11) #1
3. Determine location of CDS. (0/0)
4. Archiving and aggregation of clinical decisions. (1/0)

KEY: (# who considered action important/# who considered action feasible)

4. Controlled vocabulary for clinical activities. (0/0)
5. Controlled vocabulary for phenotypes ontology. (6/1) #6
   * Inventory of existing ontology
6. What information should face patient and how should this be organized. (1/0)
7. Define necessary federated databases needed to implement GM (EVS, Clinvar, ClinGen, Decipher, COSMIC). (0/8) #4

KEY: (# who considered action important/# who considered action feasible)

8. Define different needs for germline vs. somatic variation. (2/3)
9. Study existing solutions to ID solutions that are more robust and generalizable (variant databases, meta-databases, storage of .vcf files, informatics pipelines). (4/7) #2
10. Collection and aggregation of patient-level data. (0/0)
11. Collection and aggregation of gene/variant data (e.g. EVS, HEMD). (8/0) #4

KEY: (# who considered action important/# who considered action feasible)

12. Aggregation/cleaning house genomic medicine implementation guidelines. (0/7) #6
13. 'Automated' FH from EHR analyzed and pushed to clinicians. (0/0)
14. Develop global resource for actionable clinical variants. (6/4) #3

KEY: (# who considered action important/# who considered action feasible)