**Group 3: Clinical Genome Sequencing at Scale**

We are defining “clinical genomics at scale” to mean incorporating genomic testing or technology into routine clinical care

Below is a brief 5 question survey designed to determine if there are areas of consensus and prioritization for the Clinical Genome Sequencing at Scale breakout group, in preparation for the workshop session on Monday afternoon, July 28:

1. In order to determine where NHGRI can make the biggest impact we would like your thoughts on what type of investment and progress is most important in each of these four aspects of clinical genetics. (include text box for each item)
   1. Technology development (sequencing methods and platforms)
   2. Sequence data analysis (alignment, variant calling, copy number detection, etc.)
   3. Interpretation of the clinical genome sequence obtained based on existing knowledge
   4. Functional studies of variation identified in the genome
2. Please rank, in order of priority (1=highest, 4 = lowest), these areas for future NHGRI support:

\_\_\_ Technology development (sequencing methods and platforms)

\_\_\_ Sequence data analysis (alignment, variant calling, copy number, etc)

\_\_\_ Interpretation of the clinical genome sequence obtained based on existing knowledge

\_\_\_ Functional studies of variation identified in the genome

1. Please think about each of the following potential barriers to implement genomic sequencing in routine clinical care. How much of a barrier do you consider them? Please create a choice of the following 3 options: No barrier, minor barrier, major barrier)
   1. Cost of exome and genome sequencing
   2. Turn-around time of exome and genome sequencing
   3. Health insurance coverage
   4. Poor resources and guidance to support the interpretation of genetic variation
   5. Lack of physician understanding of how and when to use genomic information
   6. Lack of understanding of the role of most genes and variants in human disease
   7. Lack of defined clinical utility for most genetic information
2. What other barriers are impeding the routine use of genomics in clinical care? (text box)
3. How should NHGRI integrate variant discovery research with clinical genome sequencing? (text box)