SGSC Conference Call September 15, 2008 Draft Minutes

I. Present: A. Archibald, C. Churcher, R. Clark, K. Eversole, D. Hamernik, V, Hansford, L. Matthews, C. Rogel-Gaillard, and L. Schook.

Apologies: D. Milan

II. Action Items:

- 1) Archibald plans to go to Sanger in the near future to discuss next steps on the annotation project, how to engage the community in annotation, and plans for a second annotation workshop.
- III. Sequence Update: Clark reviewed the updates that he distributed to the group last week. A total of 15,385 clones have been selected for sequencing and sent to the pipeline. This covers about 92.9% of the physical map. There are 2,031Mb of total sequence (62.8 Mb of finished quality) from 12,310 clones. About 7,030 clones have been sequenced to the "improved/finished" stage. He estimated that about 74.1% of the genome had been sequenced. Clark reminded the group that the original proposal planned to sequence 16,500 17,000 clones to cover the entire genome (all of the minimal tiling path). Staff at Sanger are currently prioritizing chromosomes 6, 12 and X for sequencing so that ~70% coverage of these chromosomes can be obtained. Schook thanked Clark and the entire Sanger staff for their excellent progress.

Matthews asked the group if they preferred that all sequence come from the Cori 242 library or if other libraries could be used to fill in some gaps of missing sequence. Archibald said that the preference was for 1) all sequence from the Chori 242 library, 2) use the fosmid library to fill in gaps, and 3) use another BAC libraries to fill in remaining gaps.

- IV. Annotation Update: Archibald said that there was no update on the annotation project. He plans to go to Sanger in the near future to discuss next steps, how to engage the community in annotation, and plans for a second annotation workshop. He also said that all chromosomes now have the minimal amount of sequence coverage to be included in the next assembly.
- V. SNP Chip Update: Schook stated that a sufficient number of orders had been received to qualify for Tier B pricing (\$120/sample as product only or \$190/sample with services). Note that in an email message from Illumina on September 18, 2008, sufficient orders have been received to qualify for Tier C pricing (\$100/sample as product only or \$160/sample with services). The ~60K SNP sequences to be put on the chip have been finalized. About 90% of these SNP were discovered during this process. Some SNP on the chip were used from historical knowledge of polymorphisms in candidate genes. The SNP on the chip include SNP from the X and Y chromosomes as well as some mitochondrial SNP. About 500,000 new SNP were generated during this process. About 70% of the SNP have been mapped to the genome. The consortium plans to submit a manuscript describing the discovery of SNP by the reduced representation sequencing process.

The SNP chip is now in the manufacturing stage at Illumina. SNP chips are expected to be delivered to customers in October or November 2008. The consortium is now trying to assemble a ~500 animal panel to determine allele frequencies. The panel will include trios for a HapMap project and LD project. Schook thanked the consortium for their tremendous efforts over the past 6 months to meet this deadline. He also thanked the staff at Illumina for working closely with the consortium.

VI. Next Conference Call: The next call for the SGSC Steering Committee will be on Monday, October 20 at 8:00 am (Eastern; US).