

**Illumina Clinical Services Laboratory**

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Individual Genome Sequence Report

Dr. Naviaux
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Physician Name	Dr. Naviaux
Patient Sex	Male
Patient Record Number	N/A
Laboratory Tracking	PG0000021-BLD
Date Reported	January 19, 2010

	Collected	Received
Blood Sample	11/16/2009	11/17/2009
Saliva Sample	11/16/2009	11/17/2009

Results

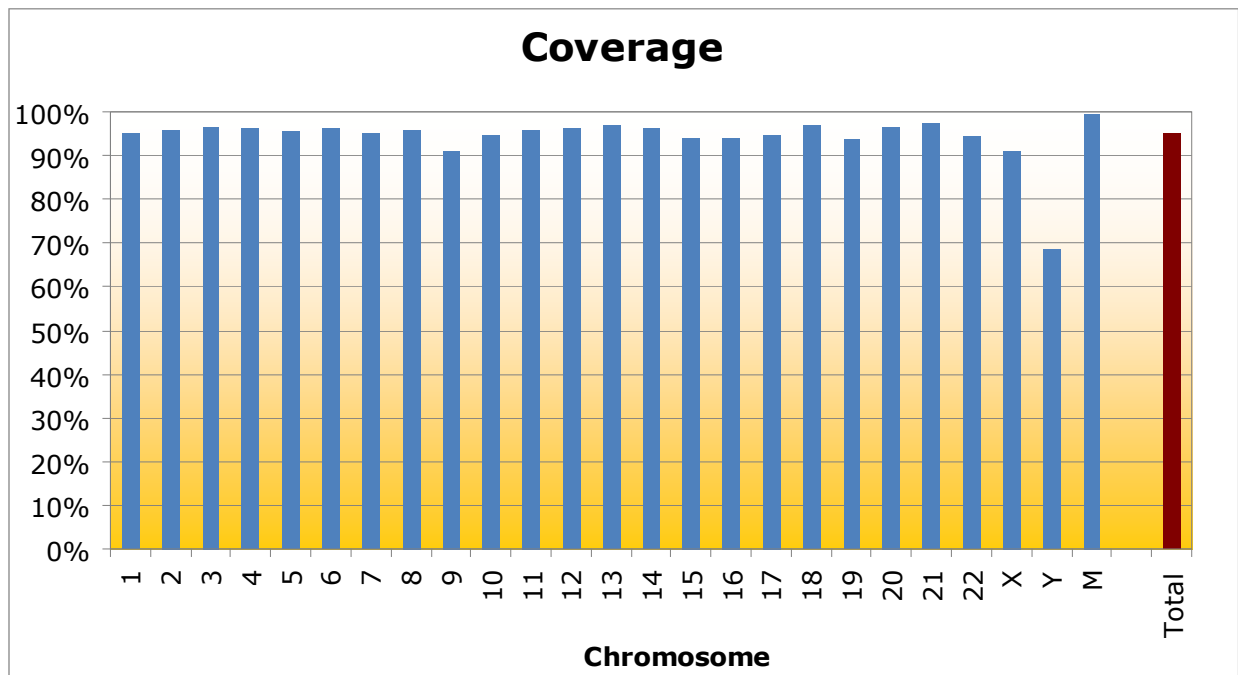
Summary Statistics	
Base pairs reported (consensus)	2,715,363,145
Percentage NCBI reference covered	95.1%
Number of SNPs detected relative to NCBI reference*	3,414,889
Number of dbSNPs genotyped*	12,809,716
Number of novel SNPs detected*	361,510
Average depth of coverage across reference genome	95.1%
Number of bases used to generate consensus	91,979,393,847

Note: The National Center for Biotechnology Information (NCBI) website (<http://www.ncbi.nlm.nih.gov/>) is a curated database sponsored by the National Institutes of Health that contains reference sequences, including the officially released builds of the human genome. The official reference build 36.1 was used to align the Personal Genome Sequence reported on here.

*The numbers reported here reflect different ways of measuring variation, and are not mutually exclusive. These numbers are further explained below.

Genomic Coverage

This graph reports on the coverage (as a percentage of the alignable NCBI published reference genome) of the patient's Individual Genome Sequence per chromosome and total. Coverage is shown by chromosome (1-22, X, Y and M, where M is mitochondria). The parts of the genome not covered here mostly comprise identical repeat sequences, which are present in the raw reads but cannot be aligned to a unique position on the reference sequence.



Single Nucleotide Polymorphisms (SNPs)

A polymorphism is a site or region of the genome that is variable. When a single base is variable, it is reported as a single nucleotide polymorphism (SNP).

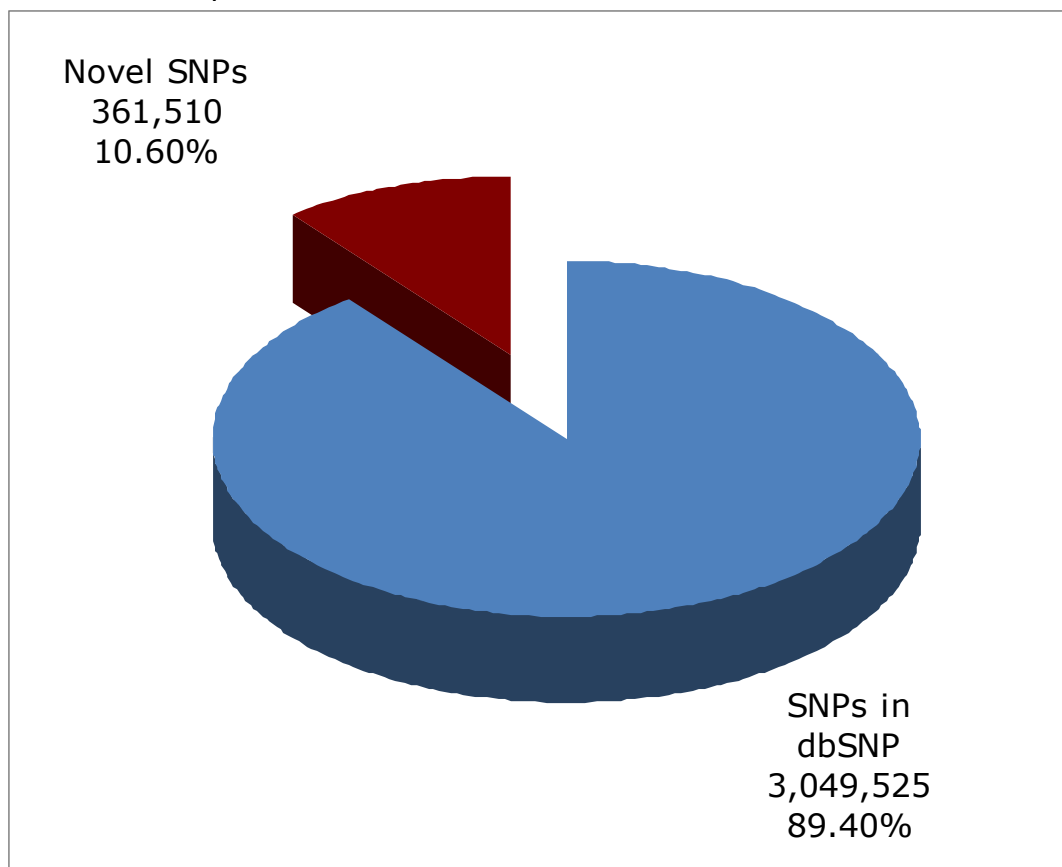
Variability can be measured by comparing the genome to the NCBI reference genome; the genome reported here was different from the NCBI reference at 3,414,889 sites.

NCBI also curates a database on single nucleotide polymorphisms (SNPs) (<http://www.ncbi.nlm.nih.gov/SNP/>). Of the more than 13 million SNPs documented in dbSNP (version 129), 12,809,716 SNPs are reported here.

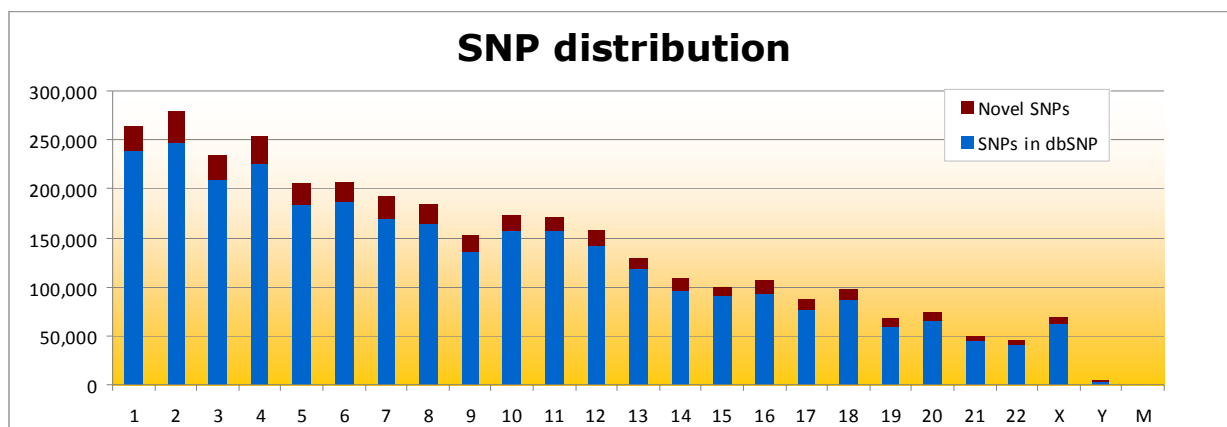
Finally, 361,510 SNPs were detected in this genome that have not been documented elsewhere. These SNPs are labeled as "Novel", however this should

not be interpreted as SNPs unique to the individual. As the public database becomes more extensive, the number of documented SNPs should increase.

For all of the following graphs, variation is reported using the NCBI reference genome as the comparison.



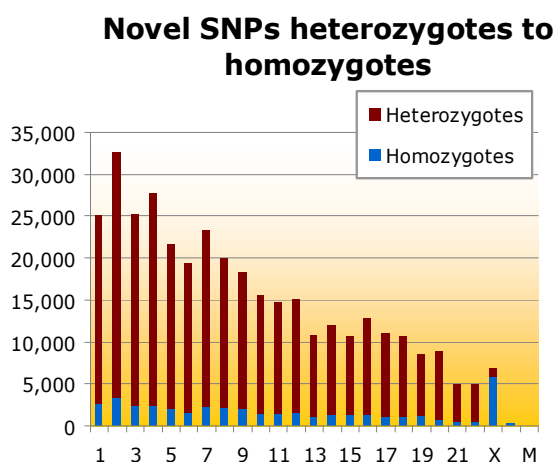
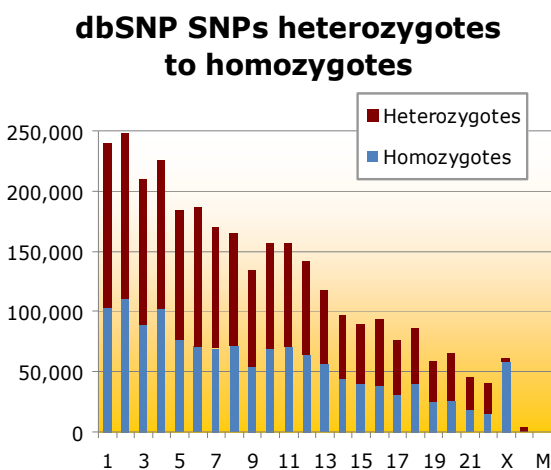
The distribution of SNPs is shown below:



Heterozygous and homozygous sites

The human genome is diploid (individuals possess two copies, one inherited from each parent) for chromosomes 1-22 (and in females, for the X chromosome as well). When the two copies differ at a particular site, the individual is said to be heterozygous for that site. The overall ratio of heterozygous to homozygous SNPs was 1.53.

The breakdown of heterozygous to homozygous sites at each chromosome is displayed below:



Methodology

Sequence was generated from DNA that was extracted from whole blood. DNA extracted from the whole blood was sequenced using the Illumina GAII sequencer. Briefly, DNA was fragmented, and the fragments were attached to the surface of a glass microscope slide. The fragments were then sequenced using fluorescently labeled nucleotides, which were excited by a laser and imaged using digital equipment.

These fragments were then assessed for quality using a variety of metrics to ensure that only robust sequences were analyzed. Fragments were aligned to the NCBI reference sequence. Fragments that aligned to more than one region of the reference genome were excluded from the report. Additionally, fragments were excluded from the analysis on the basis of quality and alignment scores. Each nucleotide site reported was sequenced an average of 30 times, so there was on average 30-fold redundancy for each base pair reported.

Illumina, Inc. made rigorous efforts to report a high quality consensus sequence, however due to a variety of factors, this genome report should not be considered complete or perfect. The regions of the genome not reported here include regions where the human reference genome has not been completely resolved, or are duplications of genetic regions make it impossible to align the fragments accurately. Although the error rates for this kind of technology are believed to be quite low, the sequence provided here cannot be considered as diagnostic. **Clinical action for any variants of potential medical concern should only be considered after further investigation confirms the presence of the variant using alternative, more focused testing specifically developed for that variant.**

This test was developed and its performance characteristics determined by Illumina Clinical Services Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration.

References

Bentley et al. (2008) *Accurate whole human genome sequencing using reversible terminator chemistry*. Nature 456:53-59.

Wheeler et al. (2008) *The complete genome of an individual by massively parallel DNA sequencing*. Nature 452:872-876.

Levy et al. (2007) *The diploid genome sequence of an individual human*. PLoS Biology 5(10):e254.

Wang et al. (2008) *The diploid genome sequence of an Asian individual*. Nature 456:60-66.

The International Human Genome Sequencing Consortium (2004) *Finishing the euchromatic sequence of the human genome*. Nature 431:931-945.

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