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Whole Human Genome Sequencing Report

Sample PG0001189-BLD

Pipeline version: v2.0.12

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Physician and Patient Information

Physician name: George Besser, D.O.
Address:
Palo Alto Medical Foundation
4050 Dublin Blvd
Dublin
CA, 94568
Patient name: K. Thomas Pickard
Patient DOB: 1963-10-08
Patient gender: M
Indication for testing: Risk Assessment

Sample Information

Sample barcode: PG0001189-BLD
Sample type: Blood - Paxgene
Sample origin: Germline
Collection date: 2013-11-20
Received date: 2013-11-21
Report date: 2013-12-07
Reference: NCBI37

SNP Assessment

Total	Ti/Tv	Het/Hom	% in dbSNP	% in Genes	% in Exons	% in Coding
3,447,773	2.08	1.61	98.79%	38.47%	1.32%	0.56%

Indel Assessment

	Total	Het/Hom	% in dbSNP	% in Genes	% in Exons	% in Coding
Insertions	134,992	1.58	85.23%	39.99%	1.16%	0.15%
Deletions	176,714	2.29	89.08%	40.23%	1.08%	0.13%

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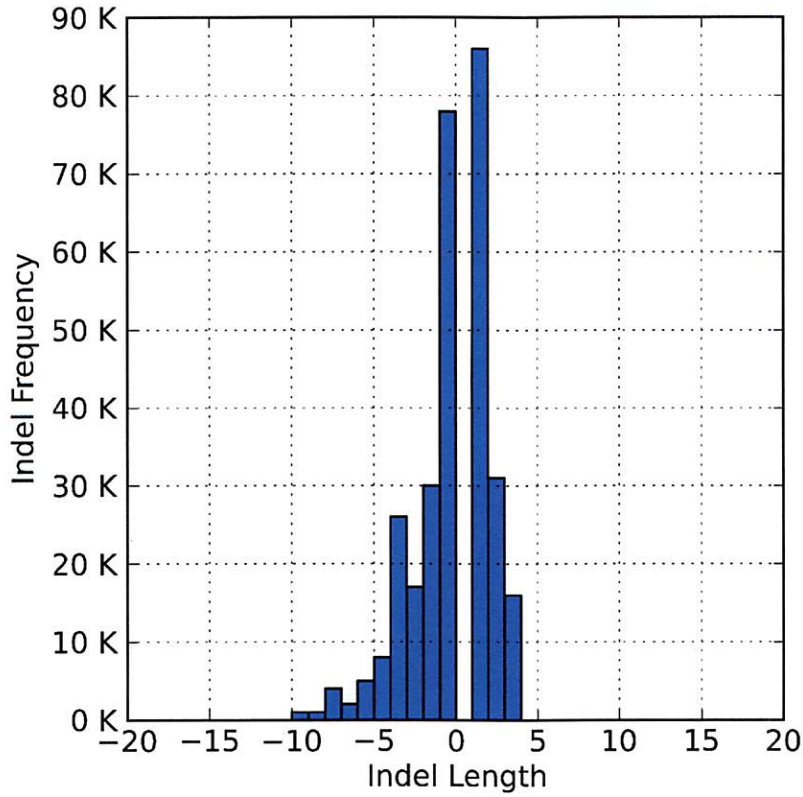
Variant Statistics

	SNVs	Deletions	Insertions
Total Number	3,447,773	176,714	134,992
Number in Genes	1,326,282	71,100	53,978
Number in Exons	45,542	1,903	1,567
Number in Coding Regions	19,204	223	199
Number in UTR	26,338	1,680	1,368
Splice Site Region	2,389	96	92
Stop Gained	79	0	0
Stop Lost	34	0	0
Frameshift	0	106	131
Non-synonymous	10,205	5	3
Synonymous	8,988	0	0

Indel Length Distribution



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Sequencing Library and Read Specifications

Below are statistics that describe the short-read sequencing library prepared from the submitted sample. Also indicated are sequencing read type and read length(s).

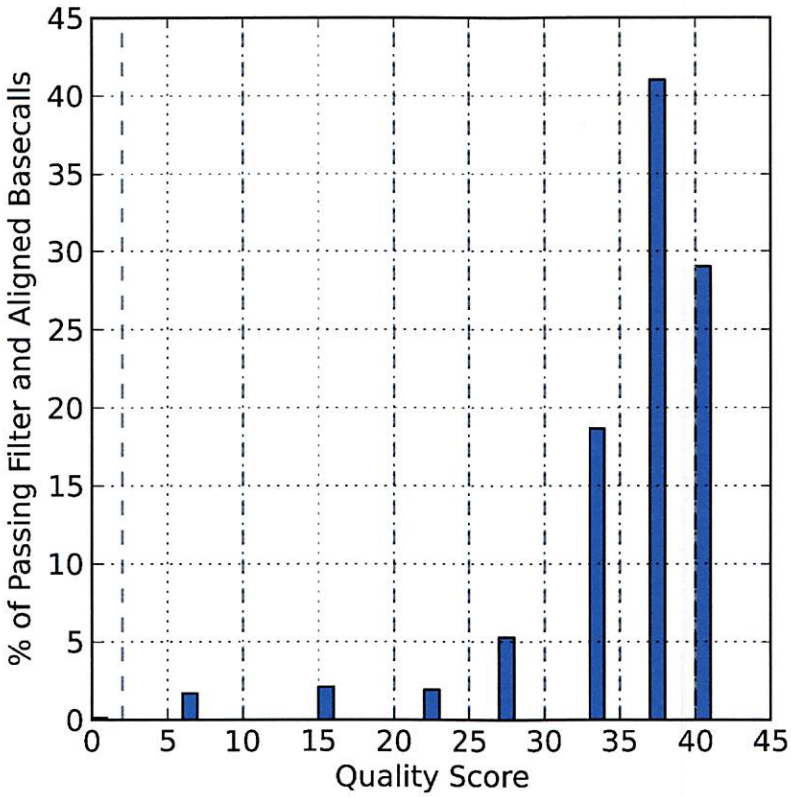
Fragment Length Median: 336
Fragment Length SD: 112
Read Type: Paired-end
Read Length: Read 1 = 100 bases, Read 2 = 100 bases

Data Volume and Quality

	Yield (Gigabases)	% Bases ≥ Q30
Passing Filter	131.99	88.04%
Passing Filter and Aligned	124.94	88.72%

The table above provides a summary of the sequencing experiment showing the total volume of bases sequenced, the fraction of bases with Phred-scale qualities greater than or equal to 30 and the fraction of bases that aligned to the reference human genome. Figure below shows the distribution of aligned basecall qualities.

Passing Filter and Aligned Basecall Quality Score Distribution



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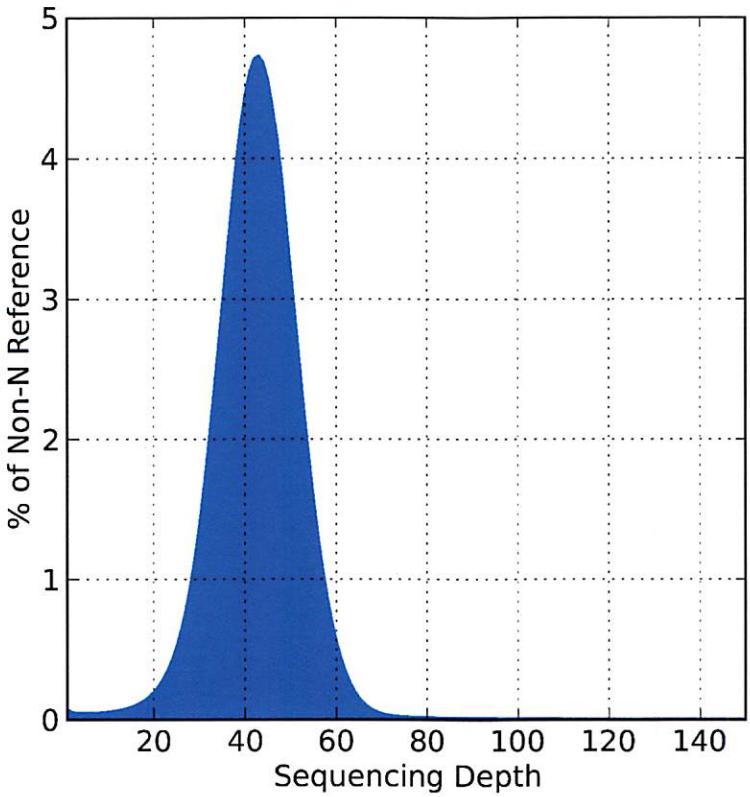
Non-N Reference Coverage Distribution

The figure and table below summarize coverage of ungapped regions of the human reference genome with unique DNA fragments. Each base in the reference genome is sequenced and mapped to the human genome an average of 30 times(x), represented by the mean depth. Also shown are the fraction of bases that are covered at or above 5x and 10x, and the fraction of bases where a genotype call was made.



	% Callable	Average Coverage	% ≥ 5x coverage	% ≥ 10x coverage	% ≥ 20x coverage
Non-N Reference	95.87%	43.08x	99.38%	99.13%	98.17%

Methodology

Sequence data were generated from extracted DNA using the Illumina HiSeq line of instruments. 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. Sequenced 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. Additionally, no positions were called when the genotype quality score was less than 30.

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

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

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

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

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

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

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