

NA19240 DATA SHEET

SEQUENCING SUMMARY

Sample	NA19240 (HapMap sample Obtained from Coriell)
Sample Source	Cell line
Gender	Female
Mapped Sequence (Gb)	187
Coverage	66X
Unique Sequence (Gb)	173
Percent of Genome (% Fully Called)	96.08%
CGI Assembly Version	1.7.0
Reference Genome	NCBI 36

VARIATION SUMMARY

Sample	NA19240
SNPs (total)	4,131,663
Novel SNPs (% vs dbSNP 129)	780,329 (19%)
SNP - Het/Hom Ratio	1.96
Small Insertions, Deletions, and multi-base substitutions (total)	593,951
Novel Small Insertions and Deletions (%)	249,621 (42%)
Small Insertions and Deletions – Het/Hom Ratio	2.04

Category	Heterozygous	Homozygous	Total
Synonymous SNPs	7870	3375	11,799
Non-synonymous SNPs	7588	3283	11,404

CONCORDANCE WITH HAPMAP

	HapMap Phase I&II SNPs	HapMap Infinium subset
# Reported	3.8M	144K
% Called	98.62%	98.48%
% Concordance	99.14%	99.86%
Homozygous Ref	99.21%	99.92%
Heterozygous	99.62%	99.83%
Homozygous Alternate	98.27%	99.80%

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support@completegenomics.com

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Published in U.S.A., January 29, 2010