Gencove's low-pass sequencing technology

gencove

What is low-pass sequencing?

Low-pass sequencing is a high-throughput and cost effective whole genome sequencing solution. The components of a low-pass sequencing assay involve multiplexing large numbers of DNA samples in a single lane or run of a sequencer, sequencing them at a very low coverage (frequently starting at 0.4x), then performing genotype imputation to make genotype calls at all sites known to be polymorphic in the population.

Gencove's imputation and analysis platform transforms the low coverage FASTQs into a VCF file with over 99% accurate variant calls across the whole genome. Low-pass sequencing returns more data and statistical power than genotyping arrays, making it the go-to technology for high-throughput genomic applications.

Benefits:

- **>99% accurate whole genome variant calls at the cost of genotyping arrays**
- High-throughput and scalable (Up to ~1500 samples per run using NovaSeq)
- Less ascertainment bias and higher GWAS discovery power than genotyping arrays
- Low DNA input required
- Easy to combine with other capture assays: Library preparation for low-pass sequencing can be combined into one workflow with target capture probes for deeper coverage at regions of interest
- Allows to perform a wide range of downstream analysis such as PRS scores or CNV analysis

Performance of low-pass sequencing vs genotyping arrays and deep sequencing

Even at very low sequencing coverages (starting at ~0.4x), Gencove's platform returns high concordance in SNP calls to the gold standard whole-genome sequencing at sites of common variation in both European and African samples.

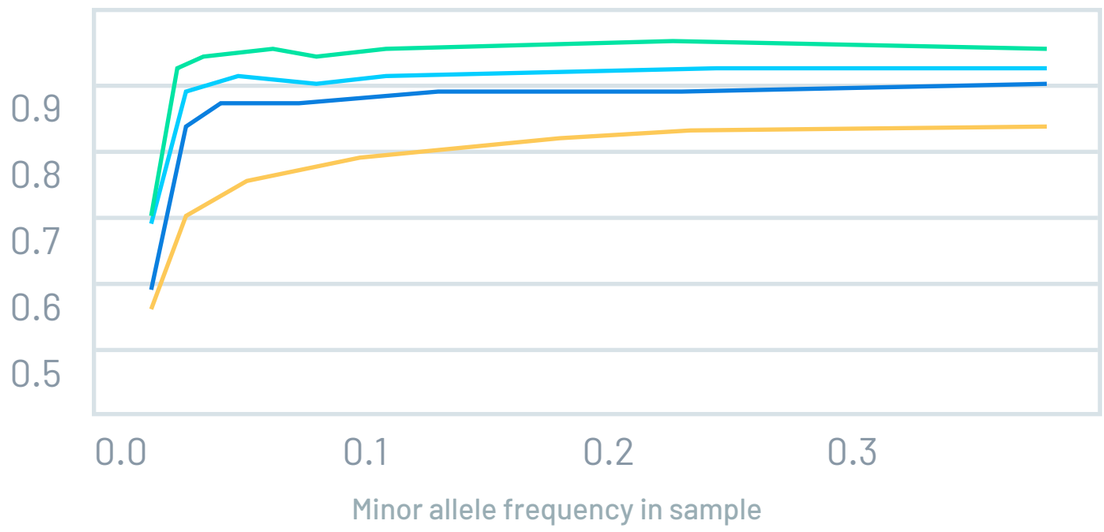
Mean overall concordance with unfiltered SNPs

	CONCORDANCE WITH WGS (%)	
EFFECTIVE COVERAGE	AFRICAN	EUROPEAN
0.4x	98.90%	99.30%
0.7x	99.00%	99.40%
1.2x	99.20%	99.50%

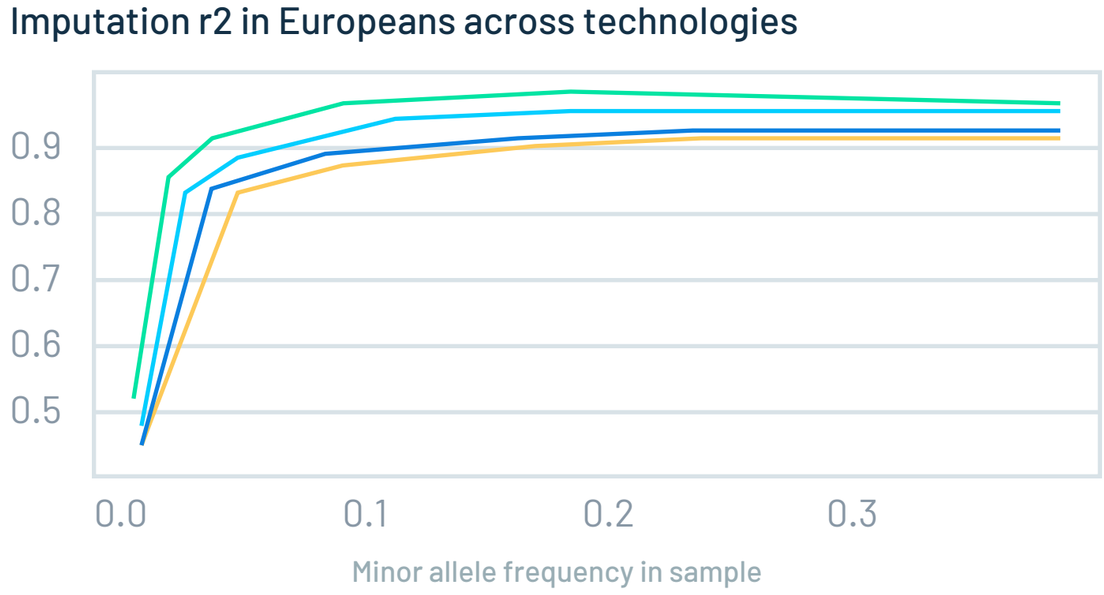
Gencove's low-pass sequencing consistently yields higher imputation quality than genotyping arrays in European and African populations, but the difference is especially pronounced in African samples.

This can be quantified with the metric imputation r^2. The higher resulting r^2 from low-pass corresponds to an effective 7% to 15% increase in GWAS discovery power for African populations and up to 6% in European populations.

Imputation r2 in Africans across technologies



Imputation r2 in Europeans across technologies



→ Results from NIH/SBIR funded validation study. Access full study here.

TRUSTED BY: Cornell University · gsk · Coriell Institute · Weizmann Institute of Science · NEOGEN · Harvard University · BGI · Stanford University

gencove

www.gencove.com
gencove@gencove.com

→ Try Gencove's SaaS and **run 5 samples for free.**