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Gencove's low-pass sequencing technology

What is low-pass sequencing?

Low-pass sequencing is a high-throughput and cost-effective whole genome sequencing solution. First, the genome is shotgun sequenced at a very low coverage (most frequently between 0.4x and 1x). The resulting FASTQ data is then uploaded to Gencove's imputation and analysis platform to obtain 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:

- **>10x** more data than genotyping arrays at a similar or lower cost
- **> 10x** reduction in cost relative to whole-genome sequencing
- **> 99%** accurate whole genome variant calls
- Low DNA input required
- High-throughput and scalable (Up to ~1500 samples per run using NovaSeq)
- Allows to **discover** new rare variants
- Less ascertainment bias than genotyping arrays
- **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

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

Gencove's low-pass sequencing at 0.5x and 1x coverage consistently yields higher imputation quality than genotyping arrays in European and African populations, but the difference is especially pronounced in African samples.

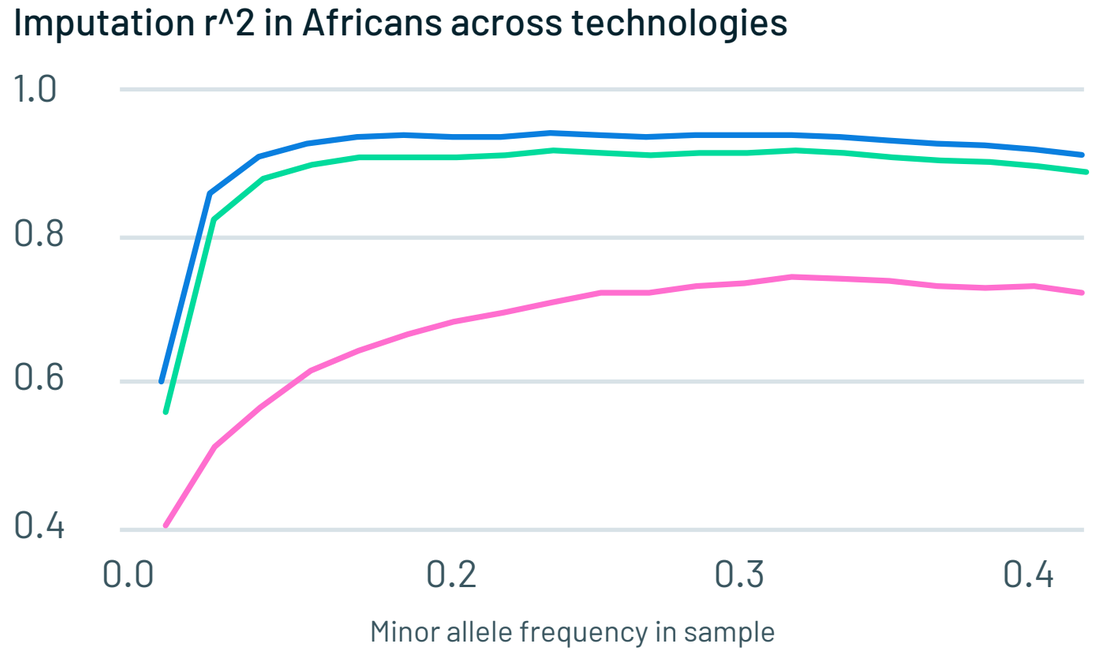

Figure 1: Performance of imputation across populations using the Illumina GSA vs. low-pass sequencing at 0.5x and 1x coverage, as measured by imputation r^2.

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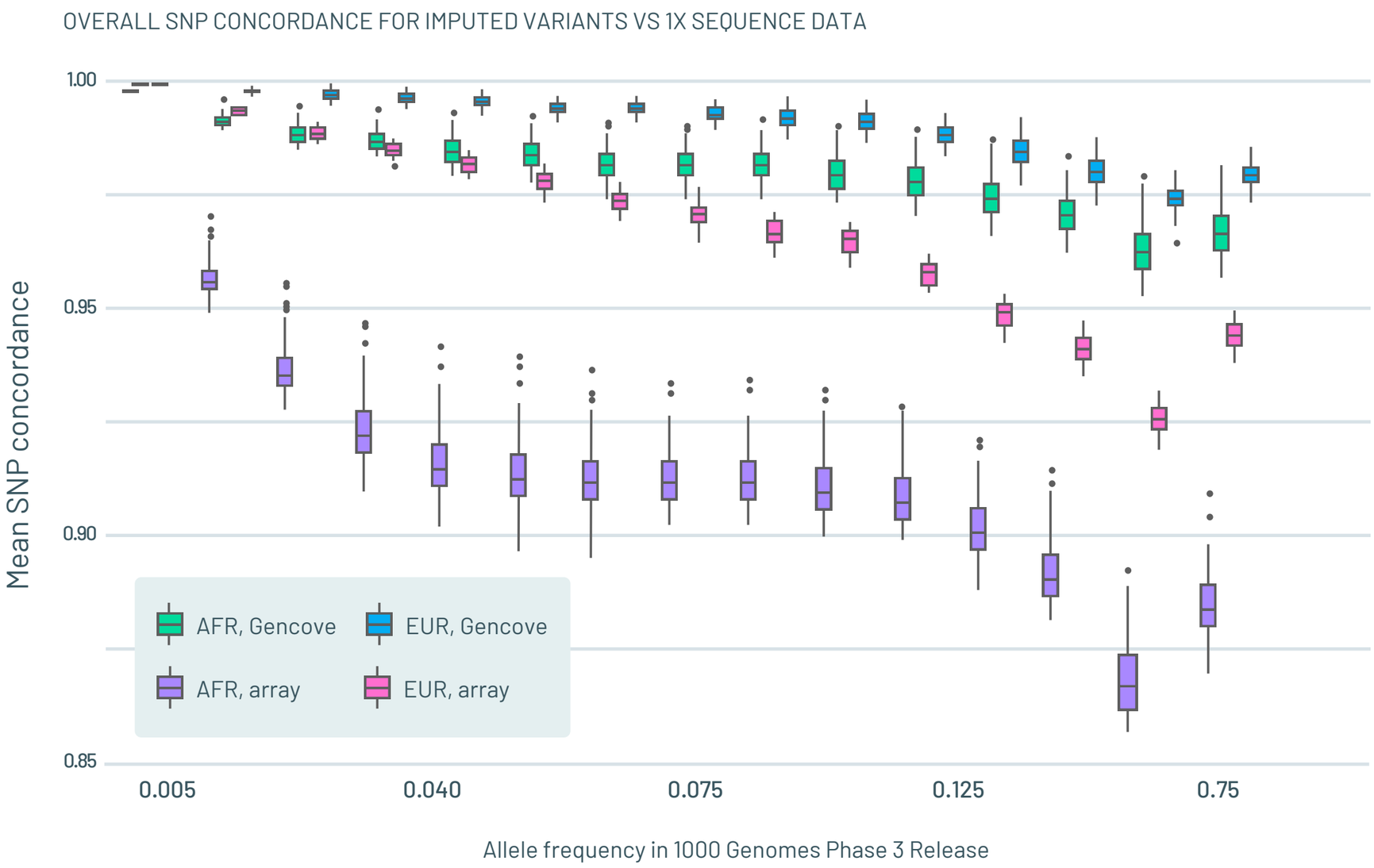

The concordance of imputed low-pass sequencing data to the gold-standard 1000 Genomes data exceeds that of imputed microarray genotypes from the Illumina GSA across all allele frequency bins and is consistently high for both African and European samples.

Figure 2: *Concordance of imputed low-pass sequencing data at 1x coverage and imputed genotypes from Illumina GSA to the gold-standard 1000 Genomes data.*

Gencove's SaaS returns high concordance in SNP calls to the gold standard whole-genome sequencing at sites of common variation in both European and African samples.

	GENCOVE'S LOW-PASS SEQUENCING DATA		IMPUTED ILLUMINA GSA	
COVERAGE	CONCORDANCE (AFRICAN)	CONCORDANCE (EUROPEAN)	CONCORDANCE (AFRICAN)	CONCORDANCE (EUROPEAN)
1X	98.80%	99.30%	91.33%	96.42%
0.5X	98.60%	99.10%		

Table 1: *SNP concordance to the 1000 Genomes Phase 3 (>1% non-reference allele frequency)*

How to do low-pass sequencing?

Library preparation

Common sample preparation methods can be optimized for high-throughput and cost-effective low-pass sequencing*

Sequencing

Perform low-pass shotgun sequencing from 0.4X and up to 6x coverage using the latest Illumina or BGI technologies.

Imputation and analysis:

Gencove SaaS transforms low-pass FASTQs into VCF files with high accurate whole genome variant calls.

**Ask us about cost-effective library preparation solutions for low-pass sequencing*

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