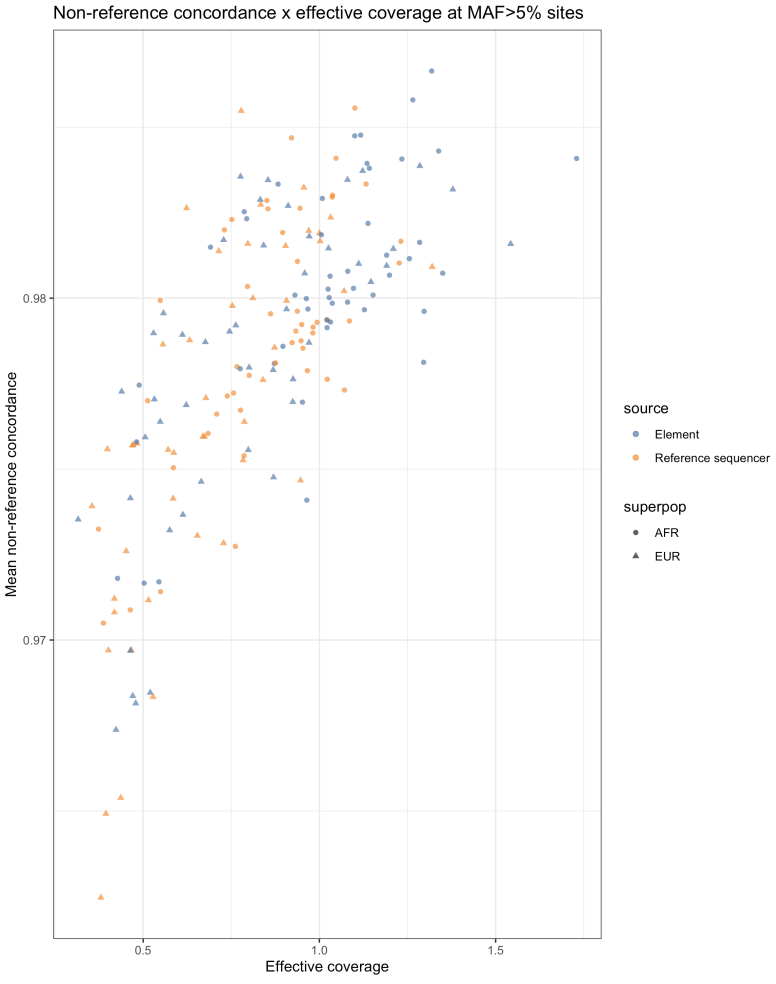
gencove

Gencove and Element Biosciences Reduce the Cost and Complexity of Obtaining Genomic Information

Gencove's low-pass whole genome sequencing and analysis platform is a hardware-agnostic high-throughput and cost-effective software layer. Gencove significantly reduces the cost per sample in three ways. First, by miniaturizing DNA requirements and library preparation. Second, by allowing customers to sequence at lower coverages and then applying the Gencove's software to impute against species-specific haplotype reference genomes. Lastly, by delivering high-quality variant calling, the Gencove platform enables ongoing downstream analysis.

To offer another option for scientists and companies seeking whole genome sequencing and analysis, Gencove evaluated the ability to process low-pass sequencing data derived from the AVITI™ System in two studies.



The AVITI System demonstrated higher effective coverage on AVITI due to a lower duplication rate.

Validation study design

The validation design mimics the Li et al. study (2021) published in Genome Research where libraries were prepared from 48 European and African samples from the 1000 Genomes Project using KAPA library prep kits. Samples were pooled and sequenced on a reference sequencer and the AVITI SystemThen the data were imputed in a leave-one-out manner to the 1000 Genomes Phase 3 reference panel.

Validation results

The AVITI System is concordant with the reference sequencer for low-pass whole genome sequencing.

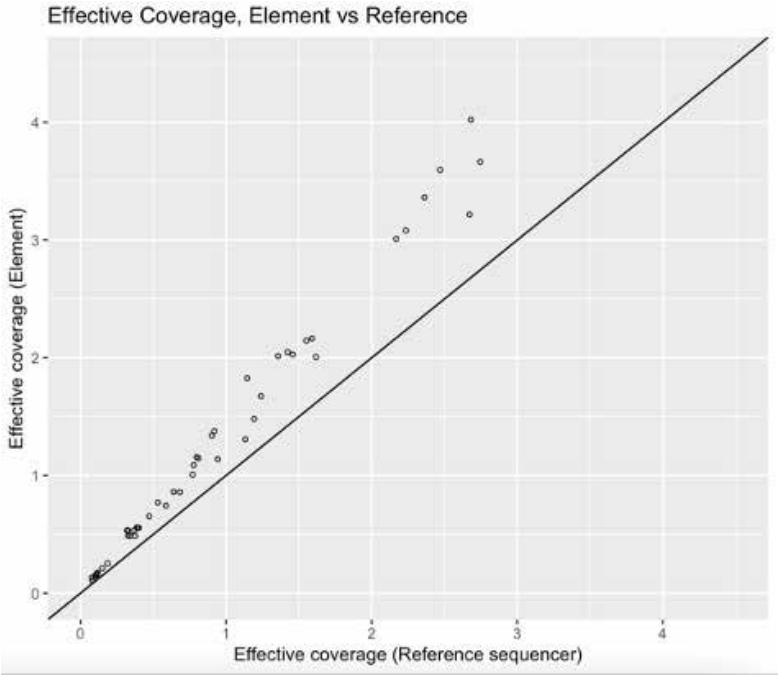
Tumor sample study

To further evaluate the AVITI System, Gencove then tested sequencing results from FFPE samples.

Tumor study design

A Gencove customer is training a model to distinguish grades of anal cancer from FFPE tumor biopsy samples. The main hypothesized source of information is in copy number profiles. We asked, “Can these copy number profiles be measured accurately with low-pass sequencing?”

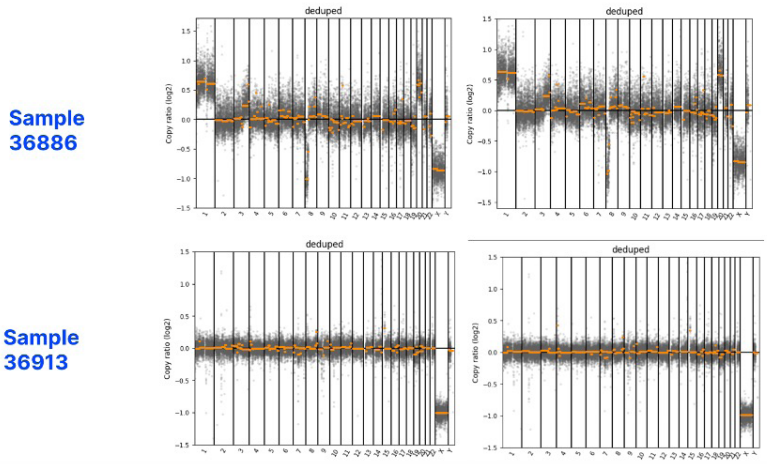
- Gencove extracted DNA, prepared libraries, and pooled
- Sent for sequencing on both the AVITI System and a reference sequencer



Tumor study results

We again found a higher effective coverage with AVITI System due to a lower duplication rate.

We also found that copy number profiles were similar across the reference sequencer and the AVITI System.



Conclusion

The AVITI System opens up additional choices for customers, and when used with the Genove platform, further reduces the cost and complexity of obtaining genomic information. Study results showed:

- Higher effective’ coverage with the AVITI System due to a lower duplication rate for low-pass whole genome sequencing.
- The Gencove platform was fully able to sequence libraries from FFPE samples with the AVITI System for profiling copy number variation.

To find out how you can use high-throughput, cost-effective whole genome sequencing and analysis for your next genomic project contact us at www.gencove.com

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