

# Low Pass Sequencing in Animals: Element Biosciences AVITI™ as a Useful Platform

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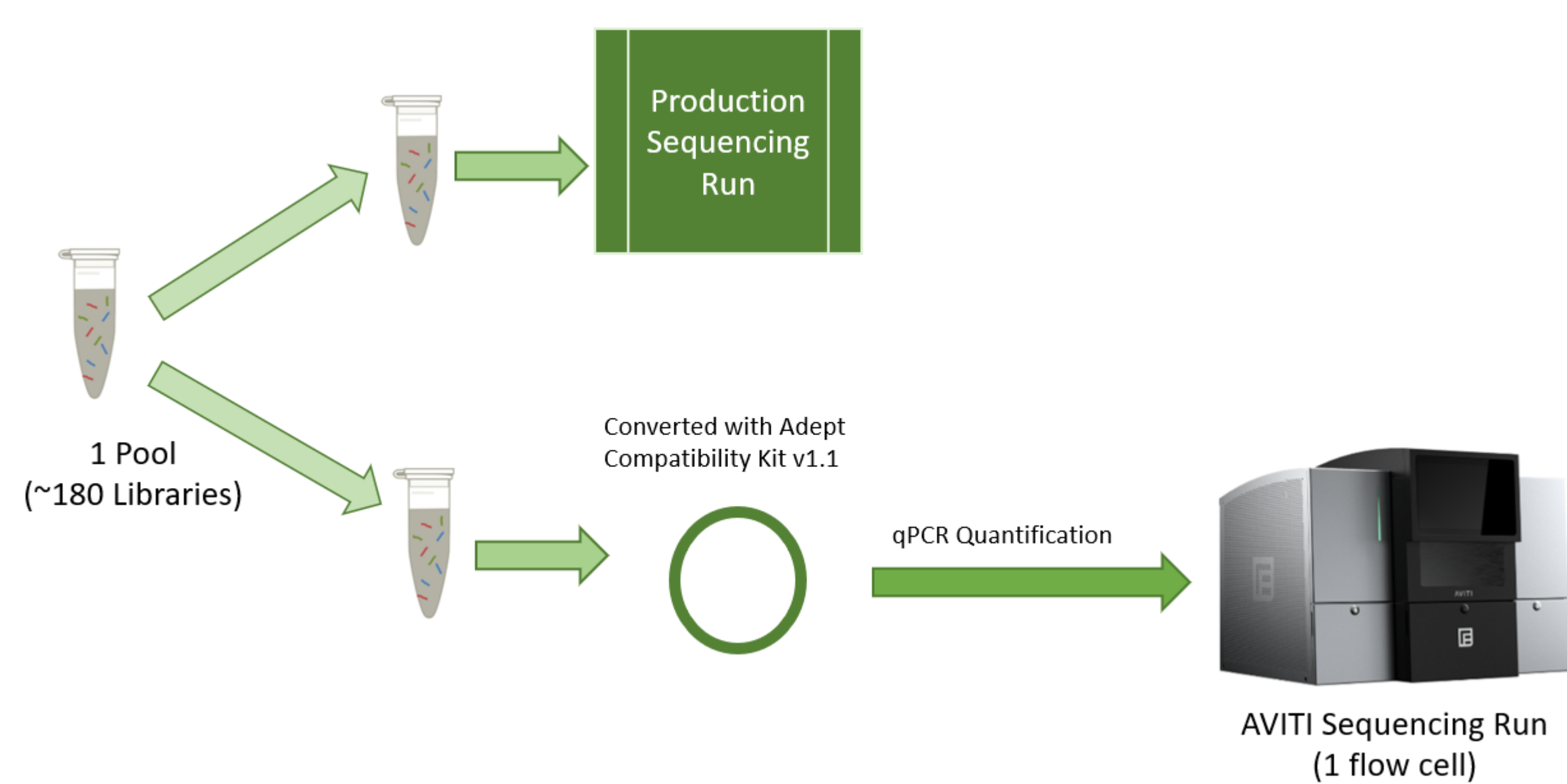
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## Abstract

Low pass sequencing combined with imputation is becoming a widely adopted tool for agricultural and companion animal applications. Combining the two yields more genotypes per dollar than other genetic screening platform. To provide high-quality data and rapid turnaround times for a multitude of applications, high throughput sequencing must be dynamic. Instead of using a production scale sequencer, which requires thousands of samples per run, we explored leveraging a benchtop sequencer which requires hundreds of samples to achieve these objectives.

In this study we compared data quality and consistency from a production scale sequencer to a benchtop sequencer. Libraries were prepared utilizing a miniaturized library preparation method, normalized, and pooled for sequencing. The library pool was divided with one portion being sequenced at Neogen Genomics, and the other processed using the Element Adept Compatibility workflow before sequencing on the Element AVITI™ System. All libraries were sequenced to a target of less than 1x coverage.

Demultiplexed fastqs were adapter trimmed, imputed using the Gencove imputation platform, and analyzed for concordance. Analysis showed comparable sequencing performance metrics between the two platforms (91% >Q30 with AVITI™ vs 90% on the production sequencer). Overall data consistency was high when compared across the two platforms with >99% concordance for imputed genotypes. Because fewer samples are needed to fill sequencing runs, faster turnaround times can be achieved with the Element AVITI™ System while maintaining data quality and consistency at a price point similar to production level sequencers.

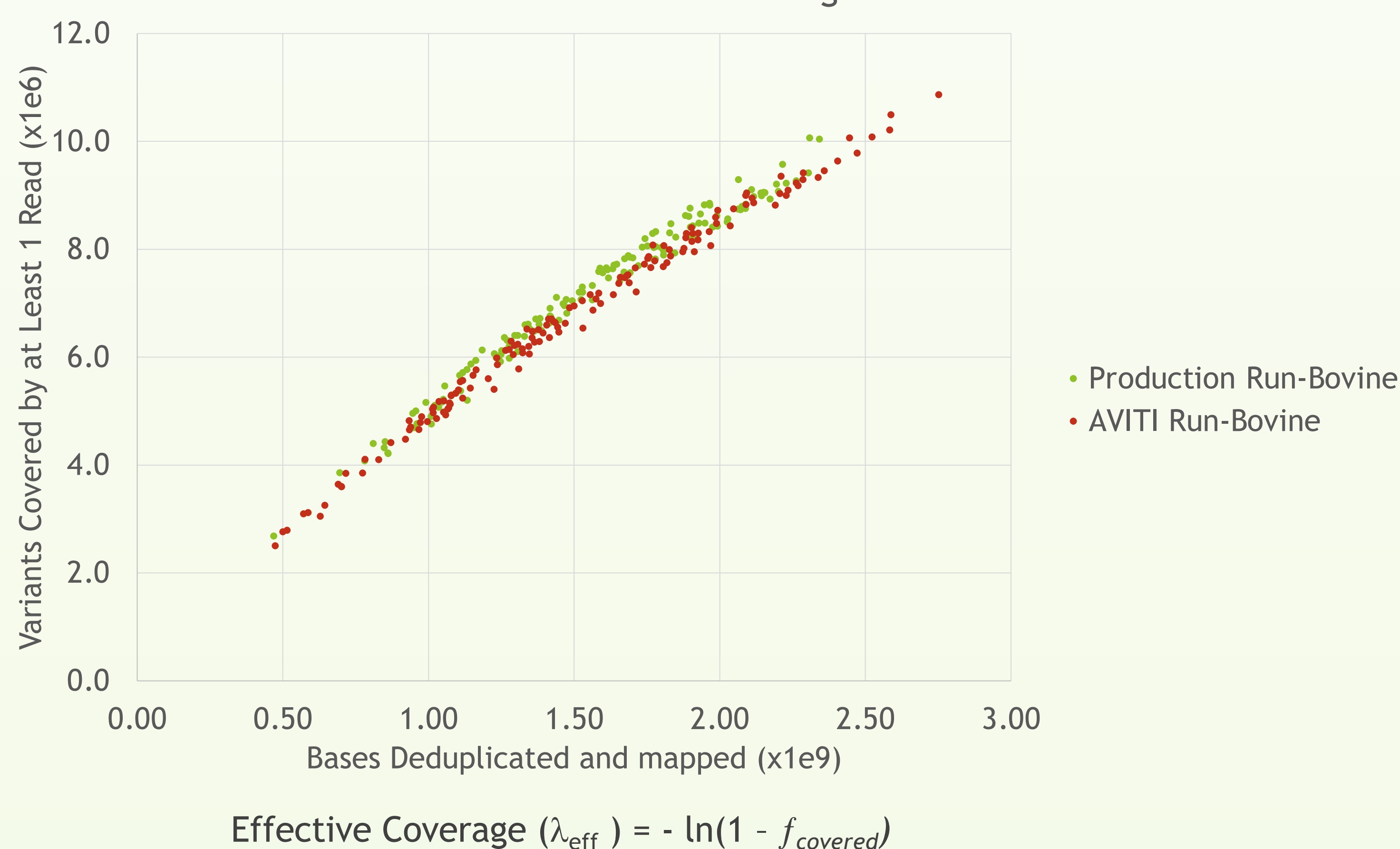


## Run Summary Statistics

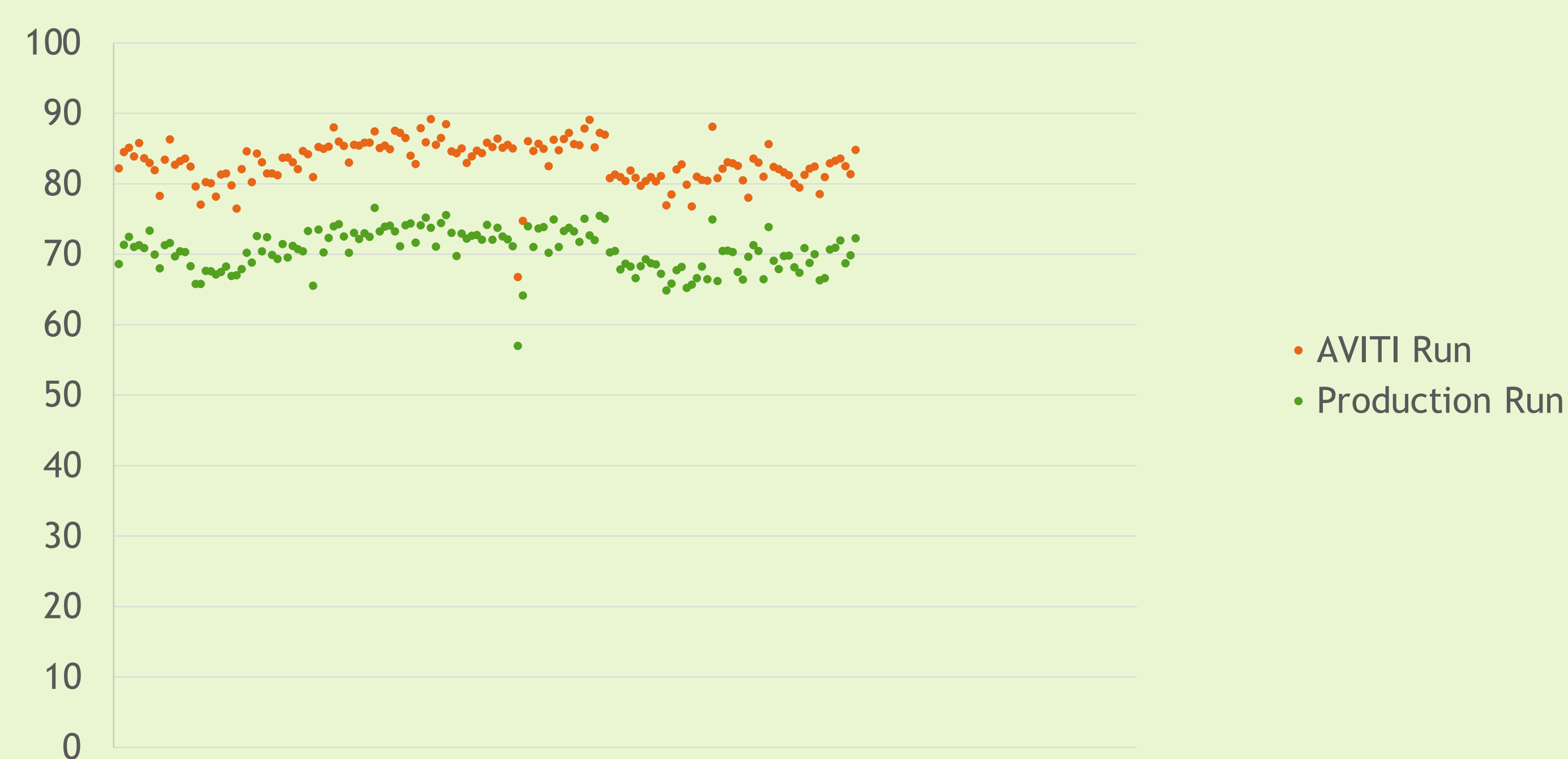
	Production	AVITI
Run Yield (Gb)	344	307
Average Estimated Coverage (x)	0.65	0.51
Average Effective Coverage (x)	0.45	0.42
Total % >Q30	90.4	91.2

## Results

### Effective Coverage



## Conversion Rates



Per Library Conversion Rates of Estimated to Effective Coverage

## Data Statistics

	Production Run	AVITI Run
Deduplication Rate	83.4%	99.5%
Mapping Rate	82.7%	99.0%
SNP Call	7,214,065	6,743,317
Effective Coverage	0.454	0.421
% >Q30	90.5	91.5

## Concordance of Imputed Genotypes

BOVG100Ki-Quality Filtered	99.5%
BOVG100Ki	95.0%
BOV770Ki	96.2%

Production Run vs AVITI Run

## Concordance of Imputation Data to Microarray Data

	Number of Markers in Common	Number of Correlated Genotypes	Concordance
Production Run	88,565	83,610	94.4%
AVITI Run	88,565	83,373	94.1%
Production Run-Quality Filtered	74,034	71,495	96.5%
AVITI Run-Quality Filtered	72,737	70,198	96.5%

Comparison of GGP100K array genotypes and imputed genotypes from SkimSEEK™ libraries

## Conclusions

- ❖ Higher % > Q30 when compared to Production Run
- ❖ Correlation of >99% across the two platforms
- ❖ Imputation accuracy of >96%
- ❖ Lower duplication rates yield more usable data
- ❖ Reagent cost per Gb is comparable to production scale sequencing reagents making AVITI a viable alternative

## References

Low-pass sequencing plus imputation using avidity sequencing displays comparable imputation accuracy to sequencing by synthesis while reducing duplicates. JH Li, K Findley, J Pickrell, K Blease, J Zhao, S Kruglyak

