Synthetic Genomics Data Generation and Evaluation for the Use Case of Benchmarking Somatic Variant Calling Algorithms

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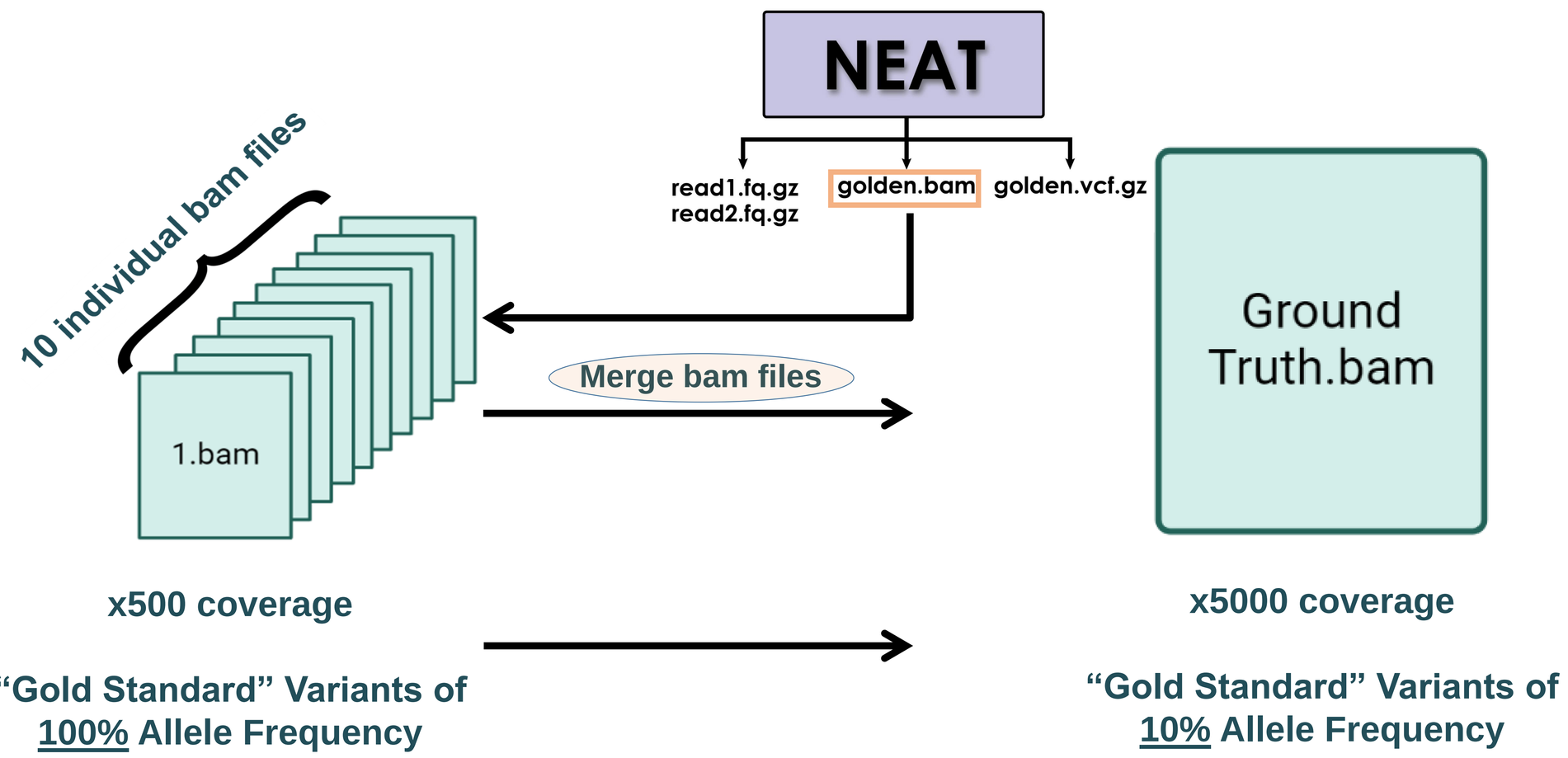
1 Synthetic «Gold Standard» Dataset Generation

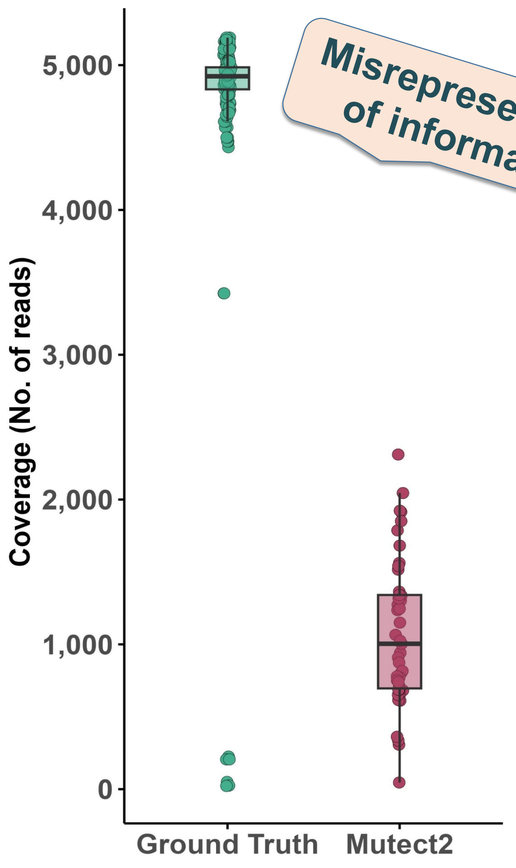
Highlights

- Generation of **synthetic genomics** data based on *TP53* gene
- Define **«Ground Truth»** SNIPs and INDELs in order to **benchmark** somatic variant callers
- Investigate the impact of variant callers in variants at **low frequencies**

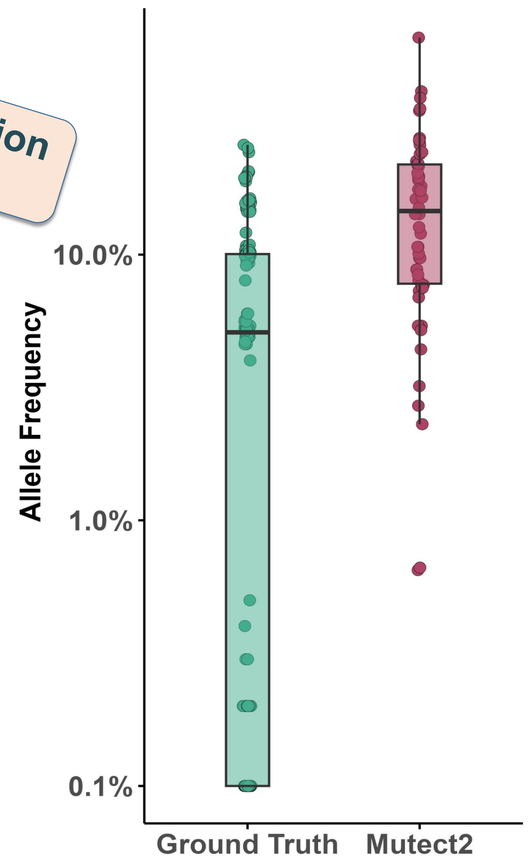


2 Benchmarking GATK-Mutect2

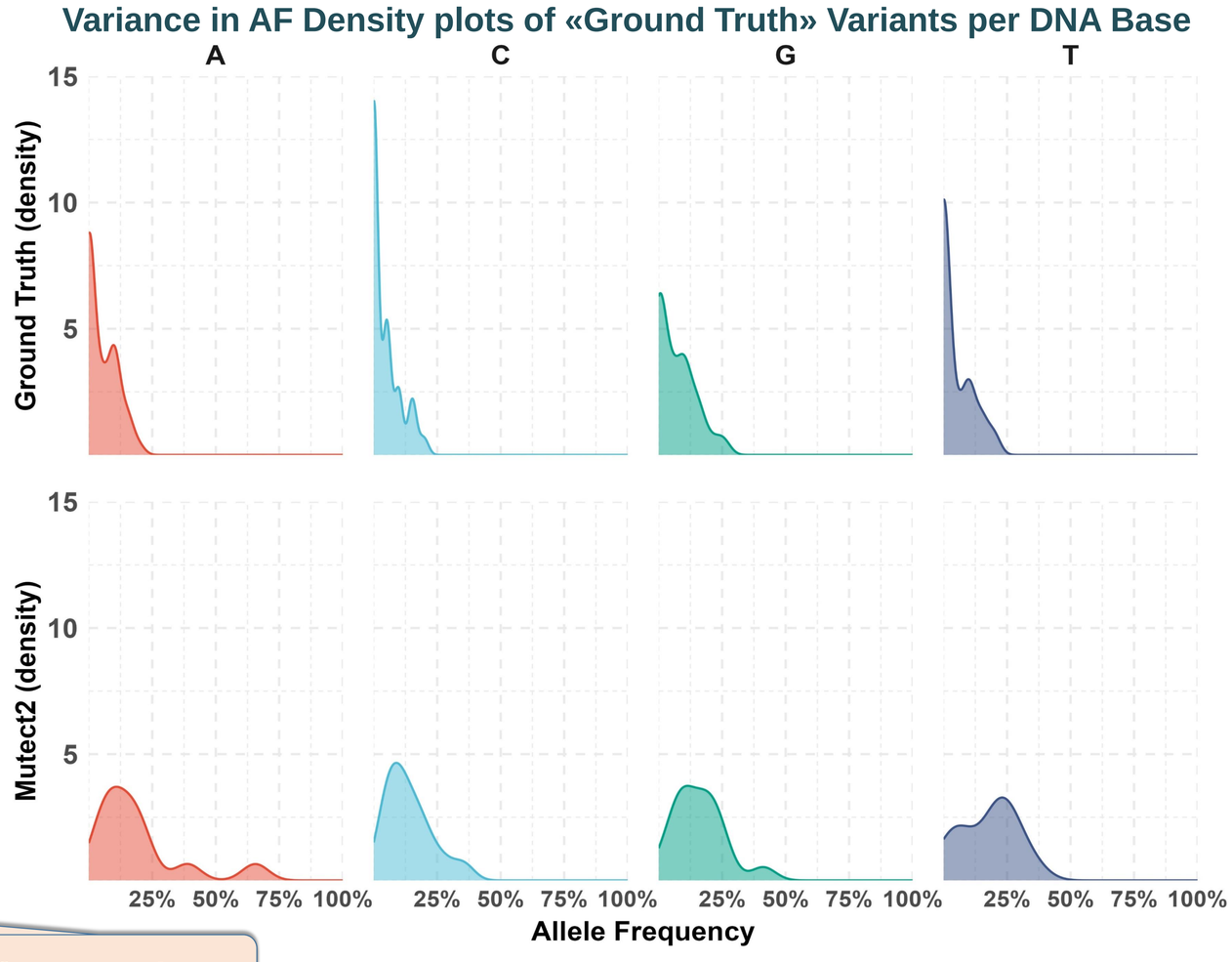
Down-sampling of coverage of «Ground Truth» Variants



Differences in AF of «Ground Truth» Variants



Variance in AF Density plots of «Ground Truth» Variants per DNA Base



Difficulty to identify variants at low frequencies

Divergence in the identification of SNIPs and their AF of «Ground Truth» Variants

Venn plot of the Overall Variants

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