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

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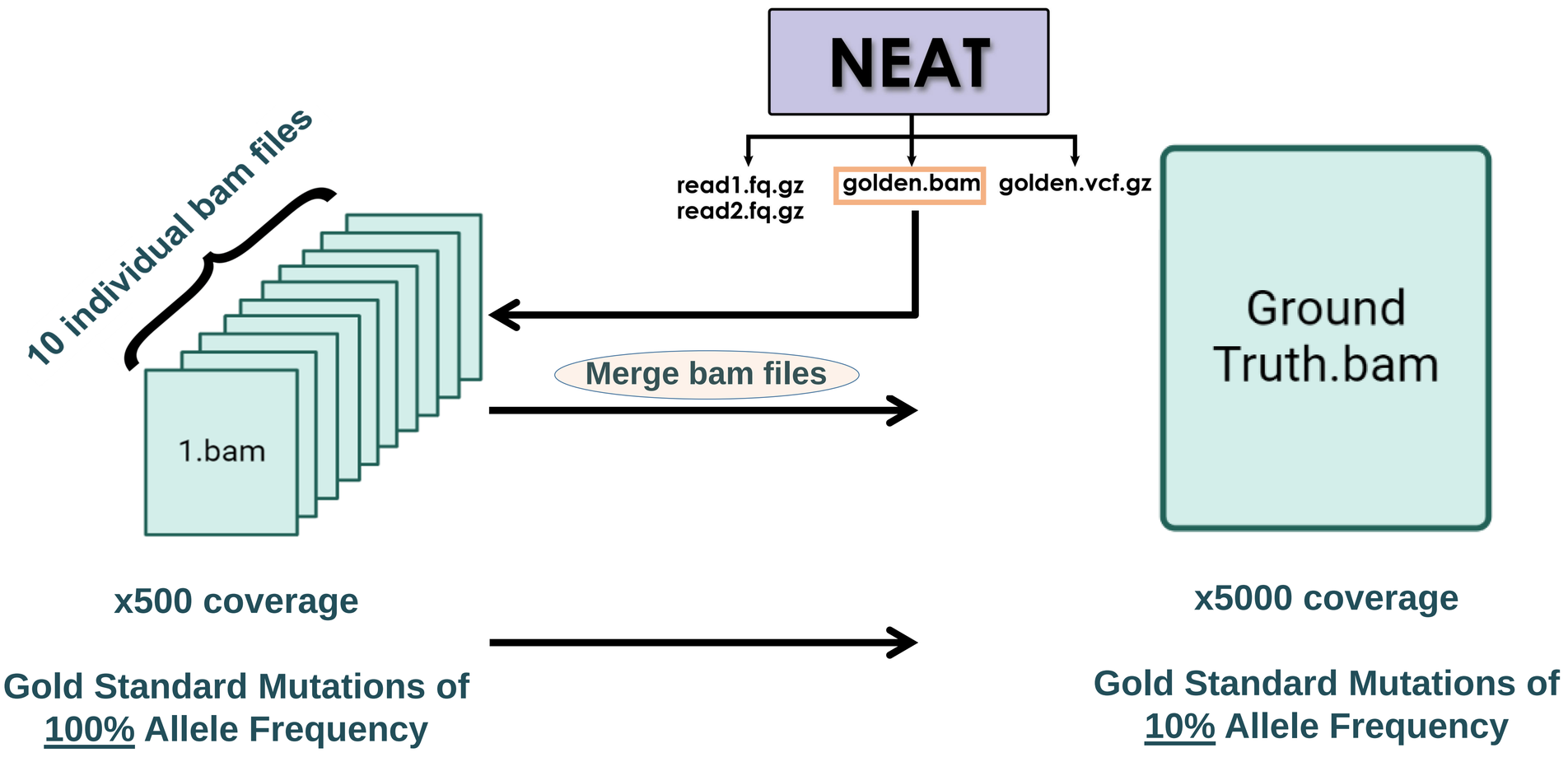
1 Synthetic «Gold Standard» Dataset Generation

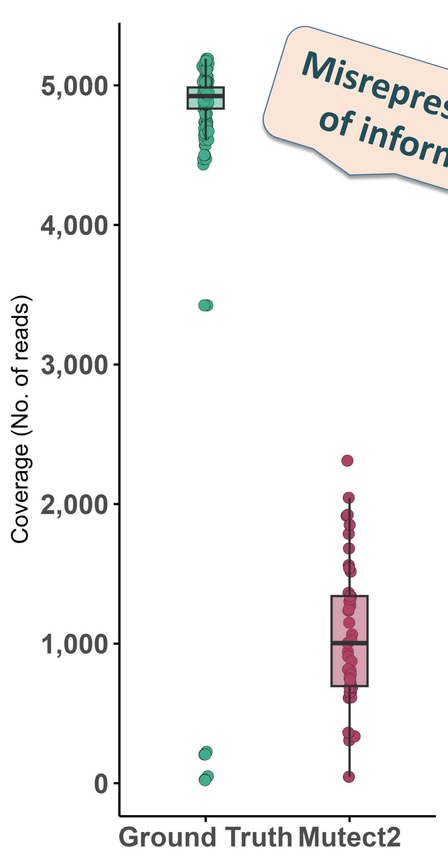
Highlights

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

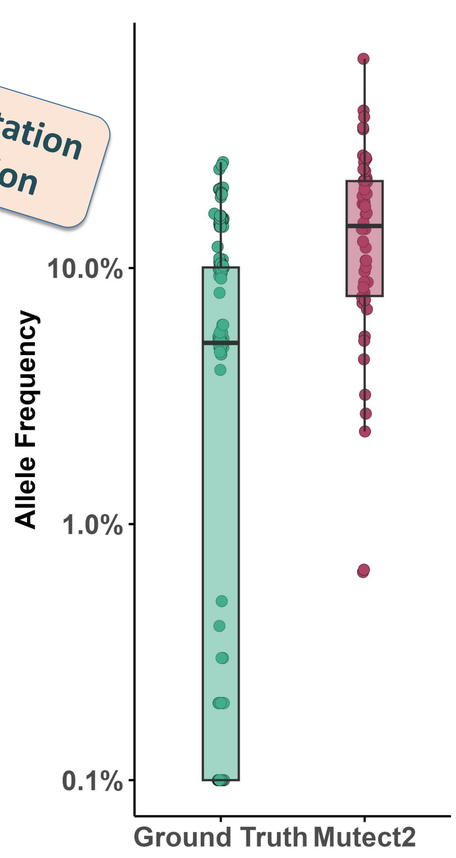


2 Benchmarking GATK-Mutect2

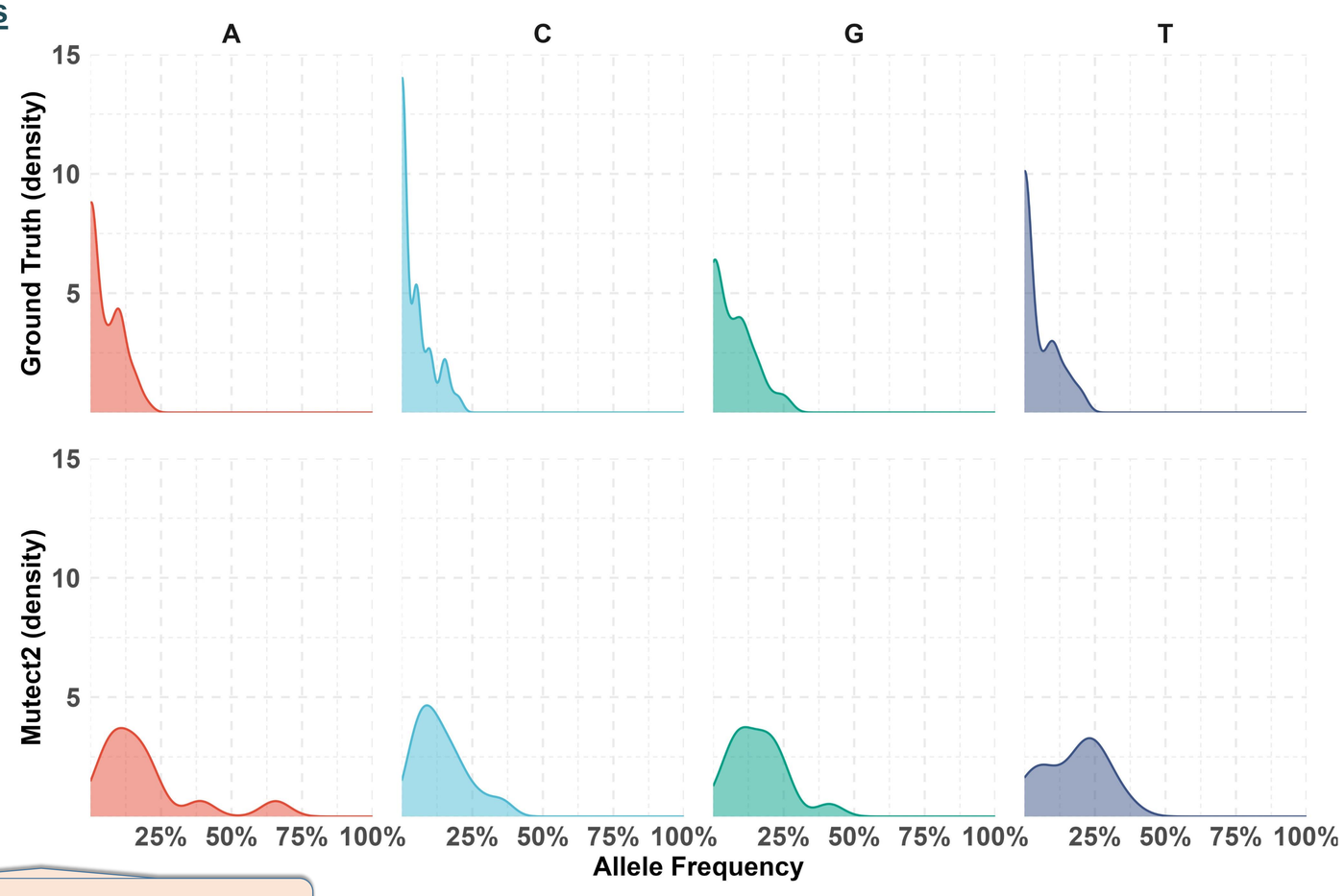
Down-sampling of coverage of «Ground Truth» Mutations



Differences in AF of «Ground Truth» Mutations



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



Difficulty to identify mutations at low frequencies

Venn plot of the Overall Mutations

Divergence in the identification of SNVs and their AF of «Ground Truth» Mutations

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