Synthetic Genomics Data Generation And Evaluation For The Use Case Of Benchmarking Somatic Variant Calling Algorithms

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MOTIVATION

- Variant calling plays an important in identifying genetic lesions.
- In the case of variants at low frequency (≤10%) identification becomes more challenging.
- The challenge that rises is the absence of a ground truth for reliable and consistent identification and benchmarking.

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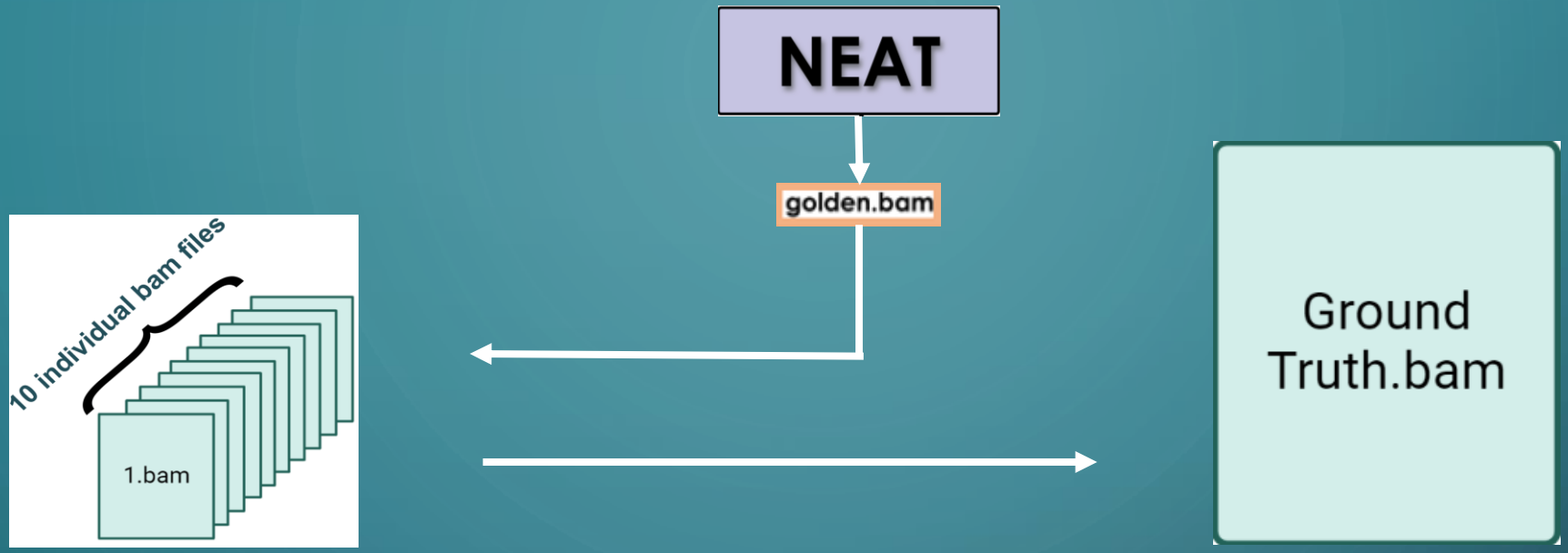
SYNTHETIC «GOLD STANDARD» DATASET GENERATION



x500 coverage

"Gold Standard" Variants of 100% Allele Frequency

x5000 coverage

"Gold Standard" Variants of 10% Allele Frequency

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COMPARING ALGORITHMS

In-silico generated dataset that contains «Ground Truth» SNIPs and INDELs

VS

Results from GATK somatic variant calling algorithm

gatk

Mutect2

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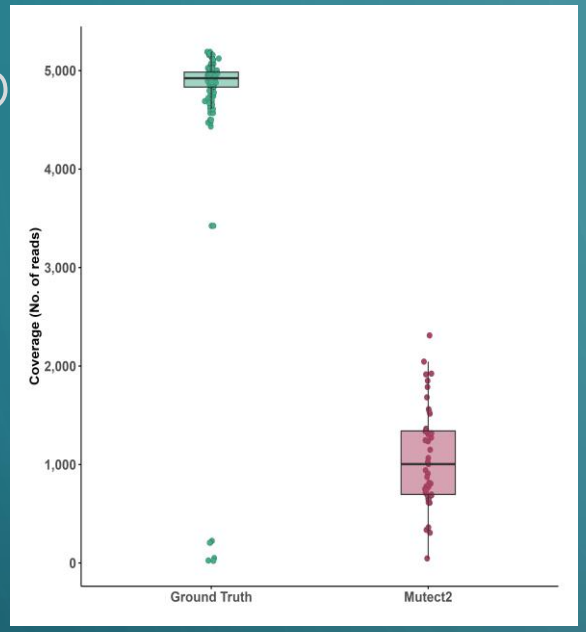
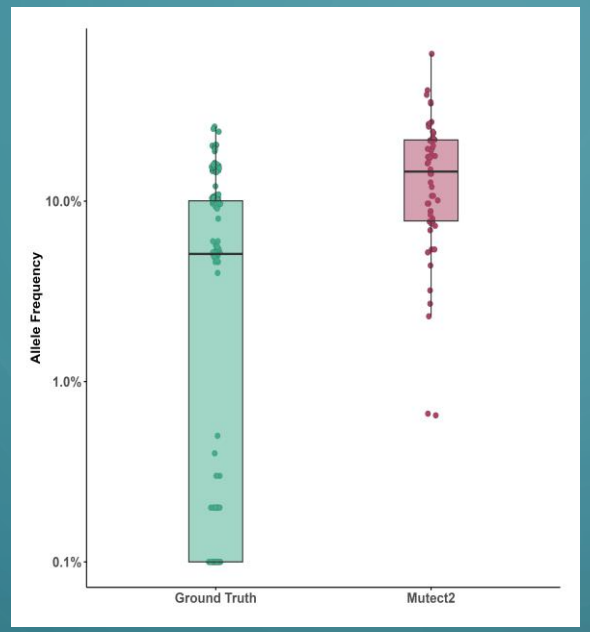
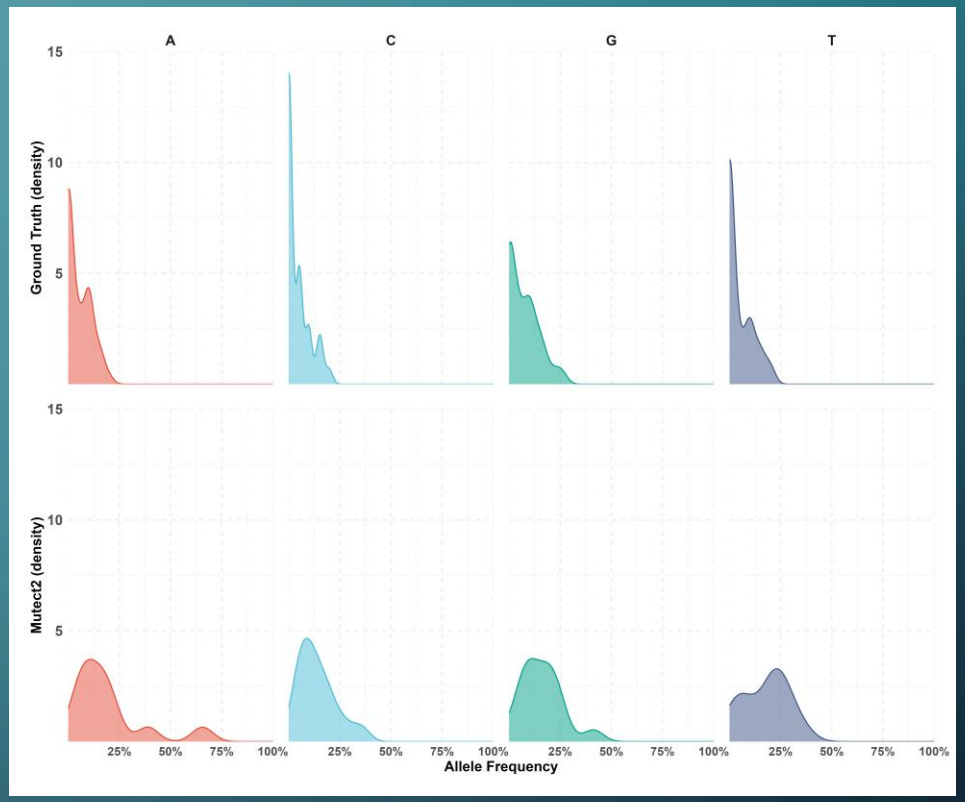
BENCHMARKING GATK-MUTECT2



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

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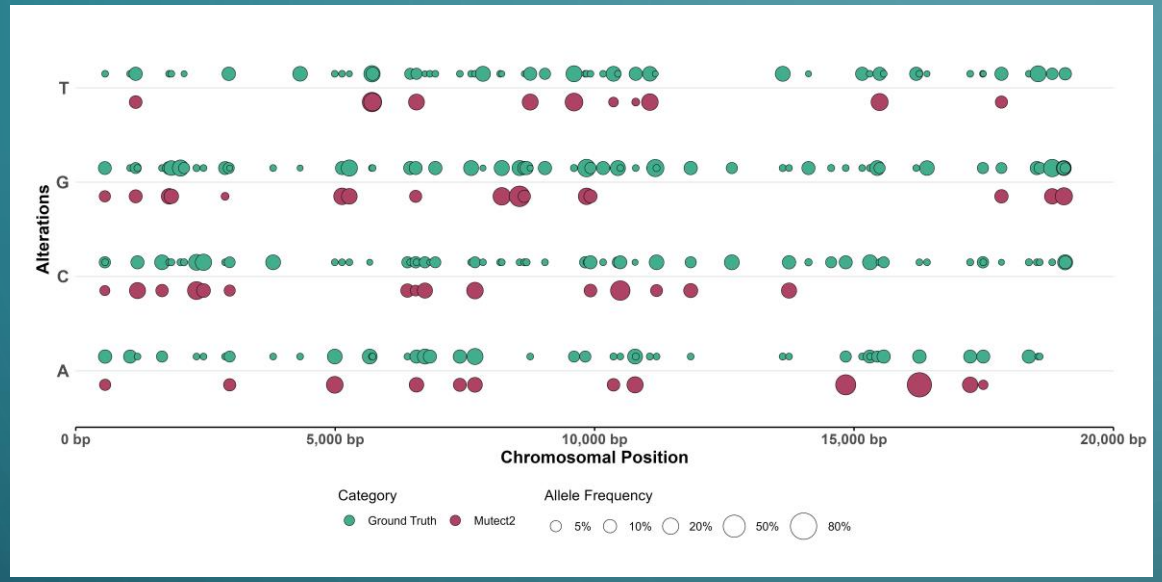
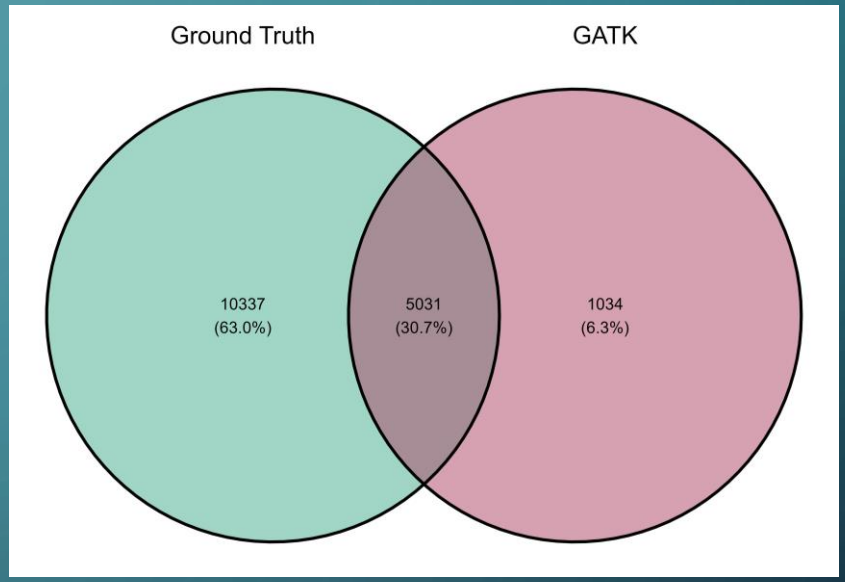
BENCHMARKING GATK-MUTECT2



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



Venn plot of the Overall Variants

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Highlights

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

To learn more about our work please visit

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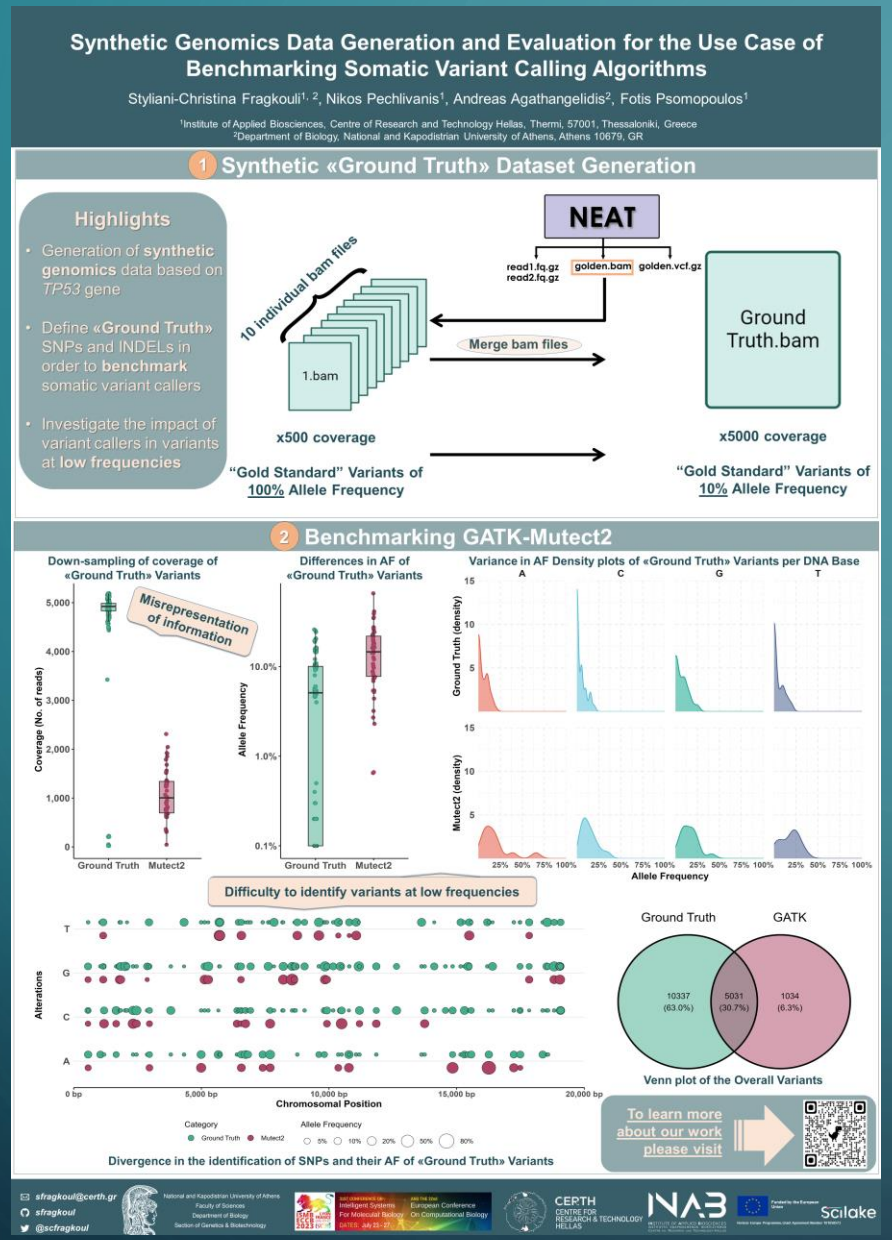

Come say Hi :)

Monday 24th of July

Poster Session A

Track: BOSC

Poster: A-102

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