

DIAGNOSTIC MEASURES FOR FUNGAL GENOMIC VARIANT CALLERS



National Human Genome Research Institute

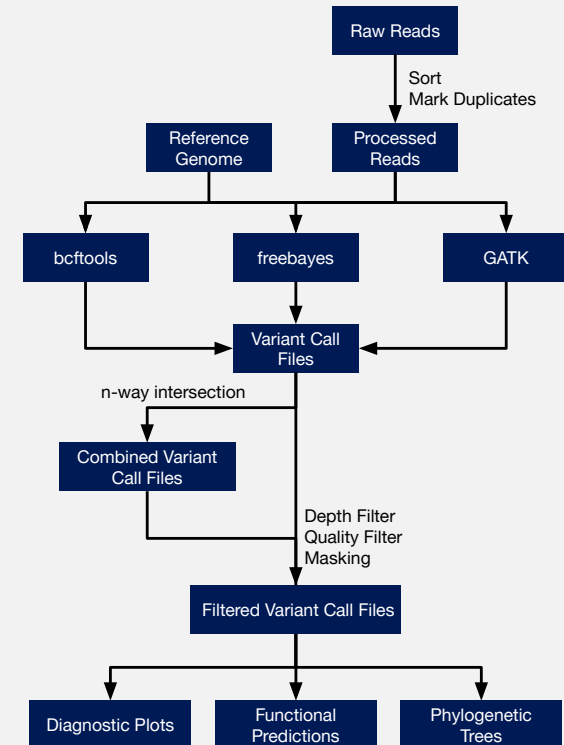
The **Forefront** of **Genomics**

Thomas Karl Atkins^{1,2}, Diana M. Proctor¹, Clay Deming¹, Qiong Chen¹, Sean P. Conlan¹, Julia A. Segre¹

¹ Microbial Genomics Section, Translational and Functional Genomics Branch, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD ² University of Minnesota-Twin Cities, Minneapolis, MN

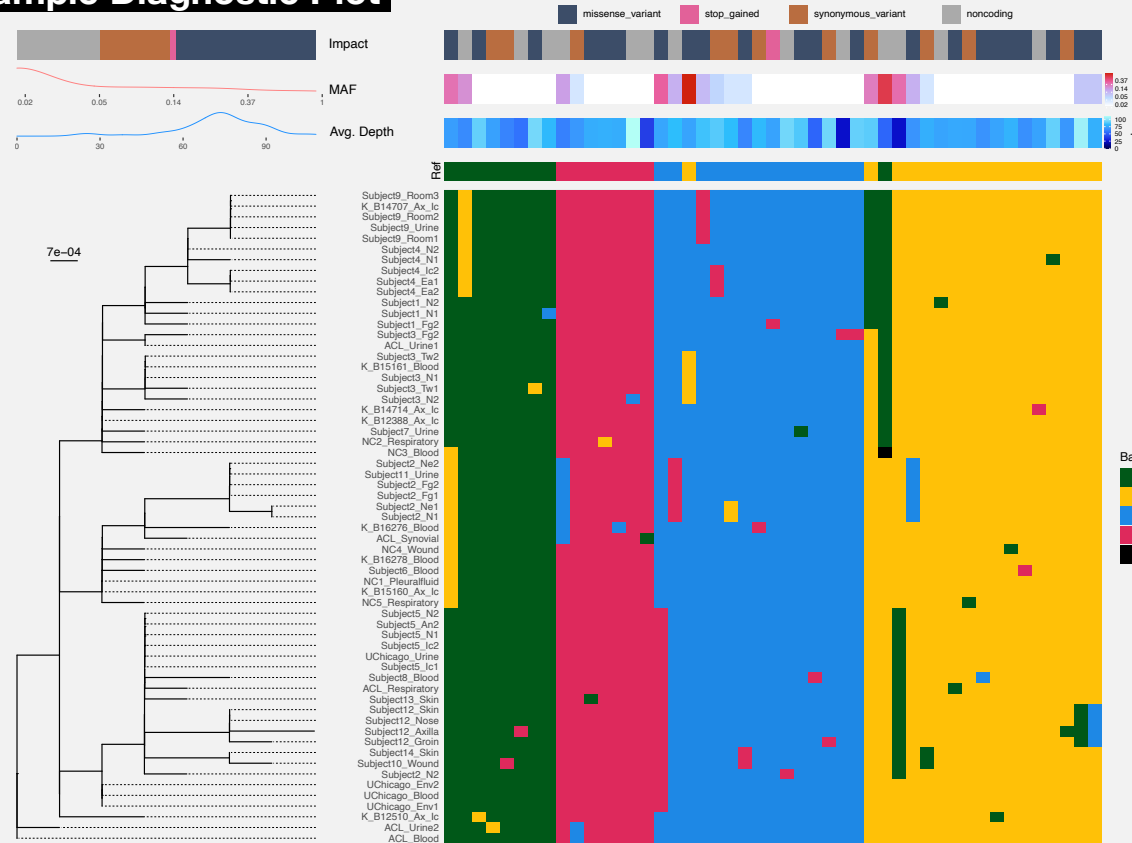
Problem: Variant callers take a set of genomic reads and return a list of genomic variants. However, output is often taken at face value without sanity checks on assumptions embedded within each tool.

Solution: We create an extensible, easy-to-use pipeline to examine combinations of three different variant callers to determine optimal model selection.



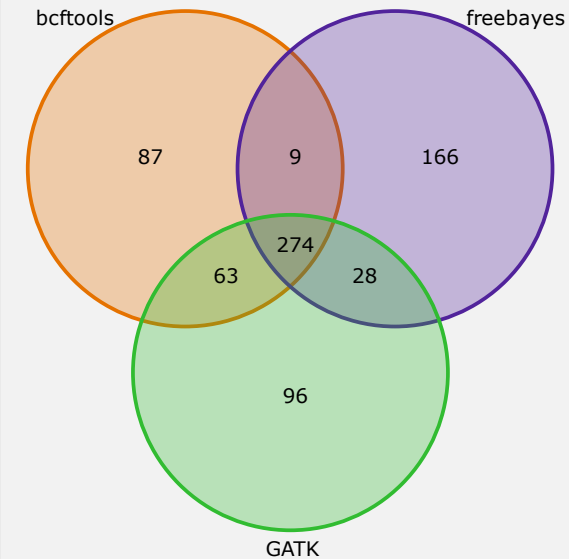
Many tools for variant calling have been developed for human genomics. These variant callers rely on **hidden assumptions that do not hold in a microbial context**. Therefore, we create a suite of diagnostic plots to **demystify and benchmark the output of these tools**.

Sample Diagnostic Plot



Impact: We test our pipeline on 60 samples of *Candida auris*, an emerging multidrug resistant fungal pathogen.

Future work: We intend to benchmark different variant callers against a set of experimentally validated SNPs to determine ground truth accuracy metrics for each variant caller in a non-human context.



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