VELSERA

The Precision Engine Company

Velsera GRAF: Building a better reference pangenome

Conventionally, reference genomes are recorded as long sequences of letters representing DNA. At locations along the sequence where multiple variations occur in a population, only a single allele is selected for the reference. This simplifies the representation, but at the expense of throwing away diverse population information.

There is a better way: build a pan-genome graph containing population information.

Best-in-class accuracy

- State-of-the-art accuracy across variant types, across WGS, WES and targeted assays
- Increased variant yield across SNPs, InDels, and structural variants, while reducing false positives

Cost-effectiveness

- Use less compute than traditional linear reference pipelines and currently established standards
- Obviate the need for costly joint calling & post-processing steps such as VSQR
- Reduce wet –lab cost by optimizing the balance between accuracy and sequencing coverage

Compatibility and no-friction deployment

- Seamless pipeline integration
- Supports standard file formats (BAM, VCF, BED...)
- Available as hosted or on-premises solutions
- User-friendly for clinical and bioinformatics users
- Integrates smoothly with existing workflows

A better approach to NGS data analysis

Ensure best in class result calling

Find more true variants

Address reference bias, increase variant yield.

Reduce false positive calls

Optimize number of variants to be assessed and reviewed.

Improved structural variant calling

Best-in-class structural variant calling with capability to call for larger SVs.

Improve time and cost efficiency

Lower your computational cost

Avoid compute-intensive post-processing quality checks because of inherent optimal reference representation.

Improve time to result

Compute-optimized algorithms with hosted and on-pre deployments.

Drop-in enhancement, no changeover

Standard file formats, compatible with your existing tools and workflows - a drop-in toolset makes for frictionless adoption.

Avoid missed insights

Represent population diversity

For samples not well represented in classical linear references, significantly increase variant yield.

Identify previously unseen variants

Optimize investment in genotyping efforts by analyzing your raw sequencing data with state-of-the-art, best-in-class secondary analysis pipeline

Collaborate with experienced science team

Collaborate with the team that pioneered this technology, build specific references, and deploy with the help of a team of experts.

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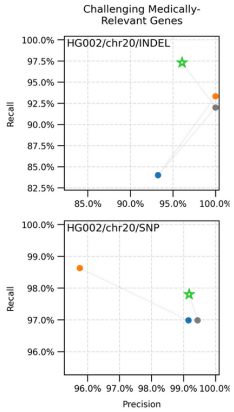
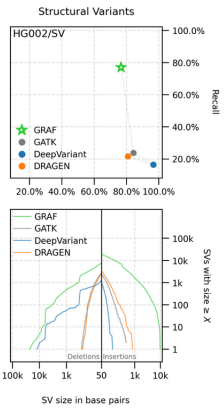
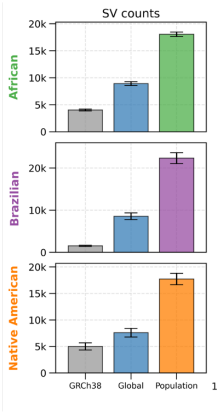
Best-in-class alignment and calling



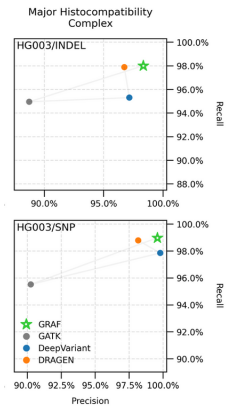
- GIAB benchmarks cover clinically relevant variants
- GRAF detects more indels in CMRG gene benchmarks
- Low false positive rate optimizes variant yield



- GRAF excels in structural variation calling.
- GRAF outperforms other pipelines focusing on short variants.
- GRAF identifies short and long structural variants accurately in one pass.



- Enriching pangenomic representation enhances SV detection and size.
- Population-specific pangenomes double SV yield.



- MHC region: highly polymorphic in human genome
- GRAF workflow excels in calling variable MHC region

Further reading

Nature Genetics:

We introduced pangenome-aware algorithms for sequencing sample analysis, showing a 0.5% increase in variant calling recall with improved read mapping sensitivity, at a lower computational cost.

Cell Genomics:

The precisionFDA Truth Challenge V2 evaluated variant calling in challenging genomic regions. GRAF excelled in accurate variant calling in the MHC region from Illumina short read samples, crucial for HLA typing.

Nature Communications:

Pangenome references encompass diverse genetic data from various human populations, aiming to enhance accuracy for non-European ancestries compared to linear references.

Manuscript Under Review

Variant calls from trio pangenome analysis improve accuracy in detecting de novo mutations, enhancing sensitivity and specificity. This automated method will help identify rare disease-associated mutations in large family cohorts.

Let's work together to apply GRAF to your data, and reveal new insights.

Contact us to learn more about GRAF and other features of the Seven Bridges Platform

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