## Secondary Analysis-Fast Alignment and Variant Calling



# A Seamless End-to-End Clinical NGS Analysis Solution, Fully Integrated with Fabric Enterprise

Fabric Enterprise is an end-to-end precision medicine platform for genomic data analysis, interpretation, and clinical reporting. The platform enables labs to easily offer a more extensive diagnostic menu of reimbursable panels, as well as support whole exome and genome tests. Fabric Genomics is a proven partner for hospital labs, commercial diagnostic labs, country genome programs, and research institutions that are implementing or scaling NGS testing.

## **Fully Automated Secondary Analysis** for Clinical Labs

Fabric Enterprise includes an optimized, best-practices alternative to BWA-GATK that allows labs to quickly launch and process NGS data with confidence. After implementation, all aspects of secondary analysis are automated, and variants are ready for interpretation using Fabric's advanced algorithms and efficient workflow. This seamless solution minimizes manual steps, enabling labs to reduce turnaround times and deliver clinical reports.

#### **Fabric Enterprise**

- Secondary Analysis (FASTQ to VCF) with Fabric Enterprise: 10x faster compared to BWA-GATK industry standard, including 20-50x faster variant calling
- Annotation and Interpretation (VCF to clinical report) - variant interpretation and reporting with Fabric Genomics' leading interpreter suite
- Lossless compression and storage of large data files (FASTQ/BAM, VCF)
- Cloud-based platform, intuitive interface, personalized training, and customer support for fast set-up and implementation

High-quality clinical results start with accurate secondary analysis and VCF files. With the inclusion of secondary analysis in Fabric Genomics' solution, customers have a single seamless platform from sequencer output to clinical report, with superb scientific accuracy and speed.



## **Faster Secondary Analysis Compared** to Industry Standard BWA-GATK

Fabric Genomics delivers a fast, accurate, and consistent secondary analysis platform using Sentieon's algorithm, winner of the 2016 Precision FDA Truth Challenge and the Precision FDA Consistency Challenge. In the Consistency Challenge, Sentieon won for top overall performance as well as for highest reproducibility. In the

Truth Challenge, Sentieon's results were very similar to the industrystandard BWA-GATK because the mathematics are identical. However, Sentieon had a performance edge because it does not down-sample reads and handles threading more robustly than BWA-GATK. Incorporation of Sentieon enables Fabric Genomics to offer 10x faster alignment and 20-50x faster variant calling compared to the industry standard BWA-GATK.



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#### **Customizable Quality Control Management**

Ensuring that samples are rigorously checked for quality is a crucial component of a robust analysis solution. Fabric Genomics' implementation team works with your lab to ensure that the quality control workflow meets your lab's guidelines. Default metrics can provide a starting point and can be customized to your needs. Samples within pre-specified bounds can be automatically passed if desired, and roles within the lab for the pass/fail QC process can be defined. An audit trail is automatically generated for these activities. QC metrics are visible at the per-sample level and also across samples. Additionally, QC metrics can be included in the clinical report output. Pileups can be viewed with the Integrated Genome Viewer (IGV) to validate individual variant quality during variant review.

### Setup, Implementation, and Customer Support

Fabric Genomics' Clinical and Implementation Services help customers set up and implement our software quickly. We have built our reputation on delivering the highest level of customer service and support.

- Our scientific team will work with you to understand your desired workflow
- We will assist in configuring Fabric platform as appropriate for the sequencing assay(s) you are running
- We will work with your lab on a QC pass/fail process that meets your needs
- We will integrate with your Electronic Medical Record (EMR) and Laboratory Information Management Systems (LIMS)

#### Secure and Cloud-Based

Fabric Enterprise is a cloud-based application that scales seamlessly as your lab grows. Fabric Genomics matches or exceeds regulatory requirements, guidelines, and standards for the privacy and security of patient data.

- Accessed via 128-bit SSL encryption; hosted in a secure data center with 24/7 monitoring
- Cloud architecture includes redundant server and backup for all
  annotated variant, report, and patient data
- HIPAA, ISO-27001, CAP, and CLIA compliance programs
- US FDA Code of Federal Regulations 21 Part 11 compliant programs for electronic signatures

#### **About Fabric Genomics**

Fabric Genomics is making genomics-driven precision medicine a reality. The company provides clinical-decision support software that enables clinical labs, hospital systems, and country-sequencing programs to gain actionable genomic insights, resulting in faster and more accurate diagnoses and reduced turnaround time. Fabric uniquely provides a comprehensive Al-based solution that spans the range of NGS applications from targeted panels to exomes and whole-genome sequencing. Fabric was the first to offer a commercial algorithm for diagnostic genomic interpretation and has partnered with leading NGS labs and major hospital systems to bring these groundbreaking advances to patients at over 50 sites running tens of thousands of tests per year. Headquartered in Oakland, California, Fabric Genomics was founded by industry veterans and innovators with a deep understanding of bioinformatics, large-scale genomics, and clinical diagnostics.

#### Learn more at www.fabricgenomics.com a demo@fabricgenomics.com 510.595.0800

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