GeneASIC NGSAAP

(Next-Generation Sequencing Acceleration Analysis Platform)

Next-generation sequencing (NGS) technologies have improved modern biological insights and hold the promise of leading the shift towards personalized medicine. However, the rapid expansion of data size has challenged computational capabilities and processing power. The amount of genomic data generated per year has reached petabytes and is expected to exceed exabytes within the next decade. Additionally, the costly infrastructure and the lack of skilled bioinformatics personnel present obstacles to promoting genetic sequencing from a rare to a routine practice. An efficient analysis tool that accurately and securely translates data into meaningful information is therefore imperative.

GeneASIC NGSAAP aims to overcome these hurdles by offering an efficient, accurate, easy-to-implement, and cost-effective on-premises solution that meets the needs of users with varying levels of expertise.

Highlights

Time-saving and accurate analysis

- Delivers around 40 results of whole genome sequencing data (WGS) with a coverage of 30X per day, achieving F1-scores of over 99%

Widely deployable device

- Easily deployed anywhere, offering server-grade performance outside server rooms

Scalable solution

- Supports the expansion of system performance for extensive data with cluster setup capability

Streamlined transmission and full control of data

- Facilitates centralized management and control of data in selected external storage resources, powered by streaming processing

Introduction

GeneASIC NGSAAP (next-generation sequencing analysis acceler

ation platform) is an ultra-fast, easy-to-use tool for secondary analysis. Relying on the reconfigurable field-programmable gate array (FPGA) technology backbone, the system significantly reduces the time required for data transmission and analysis without sacrificing accuracy.

GeneASIC NGSAAP conducts consecutive steps in the pipeline, including short-read mapping and variant calling. It recognizes FASTQ files and directly saves the results as VCF files in the specified location (**Figure 1**). In addition to its fast and accurate processing, the efficient computing architecture of the FPGA frees up host CPUs for other tasks. This allows minor tasks such as read sorting and filtering to be performed simultaneously, maximizing the throughput of sample computations.

Figure 1. The workflow of GeneASIC NGSAAP

Time-Saving Analysis for Genomics Practitioners

Conventional CPU-based systems typically take over 10 hours to process WGS data from raw reads to genotyping. However, by exploiting multiple levels of parallelism in the FPGA, GeneASIC NGSAAP can increase throughput by concurrently processing multiple samples in separate process units. Once a sample enters the calling processing, the next sample begins the mapping process in parallel (**Figure 2**). As a result, GeneASIC NGSAAP can deliver NGS data analysis results for an entire human genome at 30X coverage in approximately 30 minutes.

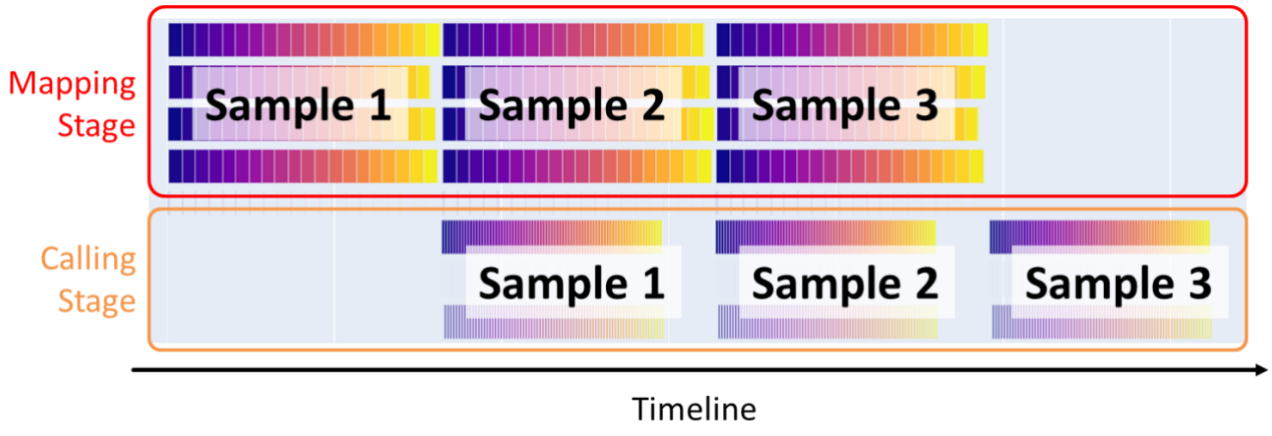

Figure 2. GeneASIC NGSAAP processes multiple samples in distinct process units concurrently

Top-Tier Accuracy

GeneASIC NGSAAP not only provides efficient secondary analysis, but also produces high-quality results. To verify the accuracy of the product, we analyzed the datasets from PrecisionFDA Truth Challenge 2016 (Table 1). With our most accurate model, controlled under Advanced Mode, the precision, recall and F1-score for both datasets were over 99%, which placed it at the same level of performance as the other top performers (>99%) in the contest.

Table 1. GeneASIC NGSAAP performance results with PrecisionFDA Truth Challenge 2016 datasets

		Precision	Recall	F1-score
HG001.WG	Basic Mode	99.77%	99.09%	99.43%
	Advanced Mode	99.74%	99.30%	99.52%
HG002.WG	Basic Mode	99.80%	98.92%	99.36%
	Advanced Mode	99.75%	99.17%	99.46%

Portable Device with Server-Class Performance

GeneASIC NGSAAP not only miniaturizes the analysis pipeline in a compact size, but also consumes low energy. It offers server-grade performance in a compact package that is suitable for use on an office floor, meeting the needs of different areas.

The rigorously designed FPGA accelerator reduces CPU resource utilization to 2%, allowing other software such as tertiary analysis to be run simultaneously. The system is compatible with standard peripherals, including USB 3.0 and local network connections, allowing it to be used in on-premises, private cloud, and public cloud environments (Figure 3). With its flexibility, GeneASIC NGSAAP can be used in various scenarios, such as hospitals, health check-up centers, labs of all sizes, and genetic service providers.

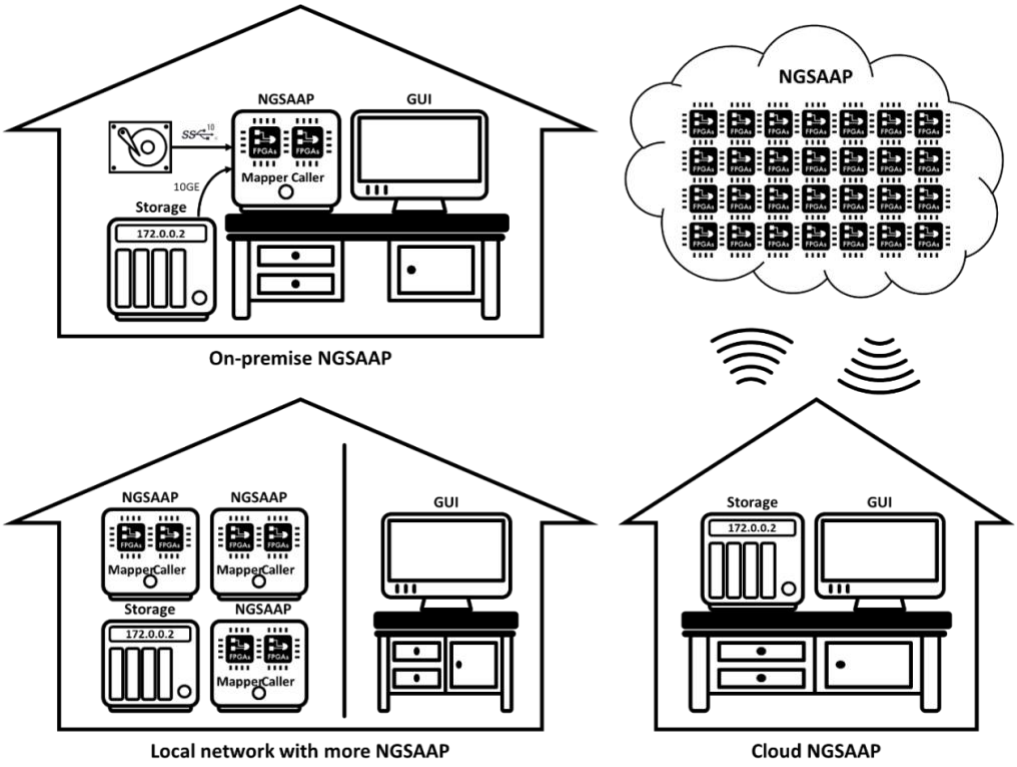

Figure 3. On-premise, private cloud, and public cloud settings with GeneASIC NGSAAP

Scalable Solution

As the demand of NGS is rapidly growing, GeneASIC NGSAAP boasts remarkable scalability, a cornerstone feature rooted in its distributed computing architecture. This design not only allows for seamless cluster setups but also makes it an ideal solution for the complex demands of genomics data analysis. As data volumes grow and computational needs intensify, the device can easily expand its capabilities through the addition of more nodes to the cluster. This flexibility ensures that performance scales linearly with the size of the computational grid, enabling researchers and analysts to process large datasets efficiently and effectively. With its robust scalability, this device stands as a powerful tool in the advancement of genomics research, offering the adaptability needed to tackle future challenges in data analysis.

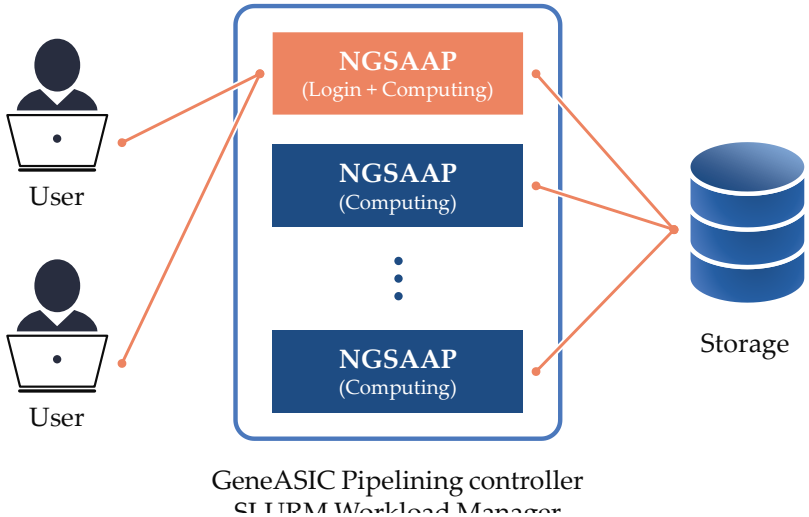

Figure 4. GeneASIC NGSAAP computing architecture

Data Security and Privacy Assurance

GeneAISC NGSAAP champions data privacy and centralized management, attributes that are significantly bolstered by its streaming processing capabilities and inherent local computing nature, particularly crucial in the handling of genomics data. Central to its design is the premise that no data needs to be permanently stored on the device itself, granting users unparalleled flexibility to designate specific destinations for both data input and output. This approach not only streamlines the flow of information but also fortifies data security, as sensitive genomics information is processed in real-time and then immediately transferred to predetermined, secure locations. By circumventing the need for internal data storage, the device mitigates risks associated with data breaches and unauthorized access, ensuring that users retain complete control over their data's privacy and integrity. This system empowers organizations to maintain a centralized overview of data flows, enhancing the management and safeguarding of genomics information while adhering to stringent data protection standards.

Learn More

To learn more about GeneASIC NGSAAP, please email info@geneasic.com

© 2024 GeneASIC Technologies Corp. All Rights Reserved. MAR 2024

6. Seamless Integration with Geneyx Analysis The Geneyx Analysis software suite transforms the high-speed output of GeneASIC NGSAAP into clinical insight. **Automated VCF Ingestion** – The VCF generated by the FPGA-accelerated pipeline opens directly in Geneyx Analysis. **Advanced AI Annotation** – Cross-referenced against a proprietary database of over 600,000 samples for superior variant interpretation accuracy. **Tertiary Analysis and Reporting** – Supports automated ACMG classification, phenotype matching, and customizable clinical report generation. **Secure Collaboration** – Enables hospitals and laboratories to manage all cases within a centralized, compliant environment (ISO 27001/27799, HIPAA, GDPR). Together, GeneASIC NGSAAP and Geneyx Analysis deliver a complete, on-premise alternative to cloud-based genomic analysis platforms like DRAGEN — faster, more affordable, and fully under your control. Learn More To learn more, contact:
GeneASIC Technologies Corp. — www.geneasic.com | info@geneasic.com
Geneyx Genomex Ltd. — www.geneyx.com | info@geneyx.com

Better Data for Better Health.