A Novel Web-Based Interface for Management of a Clinical Bioinformatics Pipeline

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The Problem

Molecular diagnostic assays utilizing Next Generation Sequencing (NGS) technologies are being rapidly adopted in a clinical setting. However, high-throughput utilization of this technology requires advanced informatics solutions that can address multiple challenges:

- Effectively organize the data and track the multiple steps in the analysis process
- Automate highly complex bioinformatics pipelines to analyze the data for multiple diagnostic assays
- Report results within a clinically relevant timeframe while ensure data integrity and regulatory compliance

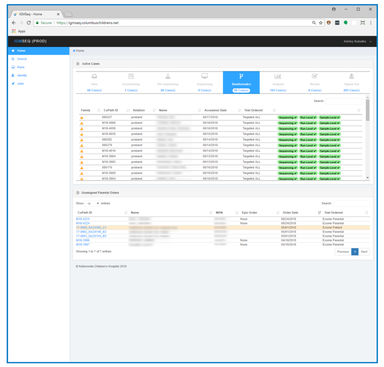
The Solution

IGMSeq is a novel web-based application that addresses multiple challenges around high-throughput clinical molecular diagnostics NGS-based assays:

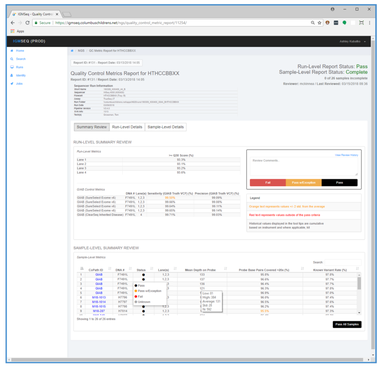
- Manages the end-to-end NGS data processing for **targeted panels**, **Whole Exome Sequencing (WES)**, and **Archer gene fusion analysis**
- **Fully automated** execution of bioinformatics pipelines
  - Sample sheet creation and BCL conversion
  - 1º analysis: run and sample level QC metrics
  - 2º analysis: FASTQ to filtered VCF (GenomeNext)
  - 3º analysis (GeneInsight) and case sign out
- **Integrates** with multiple systems and instruments:
  - Hospital-wide EMR for order receipt (EPIC)
  - Clinical reporting (SunQuest CoPath)
  - 5 models of Illumina sequencers
  - LIMS (LabVantage)
- Full **regulatory compliance** (CAP/CLIA and HIPAA)

Dashboard & QC metrics

Dashboard – review case status

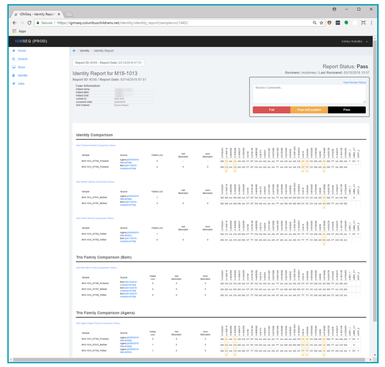


QC metric – summary level review

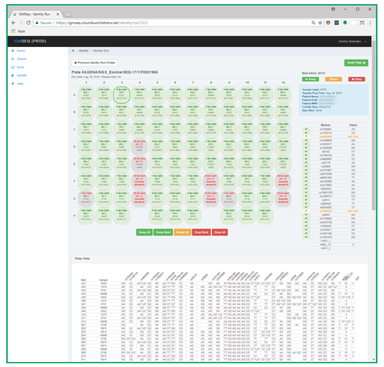


Identity Report

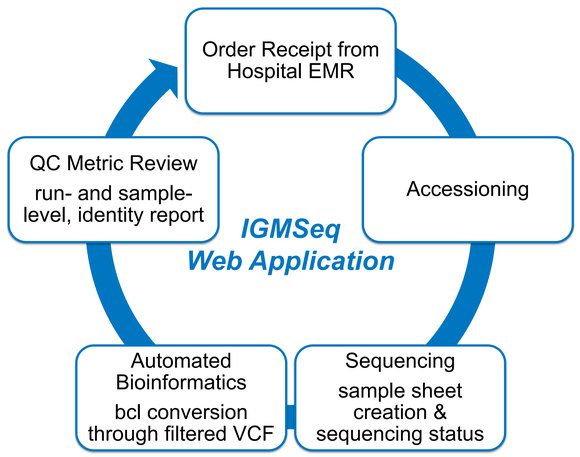
Trio identity analysis



Identity run plate review



Pipeline Process and Automation



IGMSeq has parallelized the sample-level pipeline, dramatically **reducing the computational processing time to under 30 minutes per exome**.