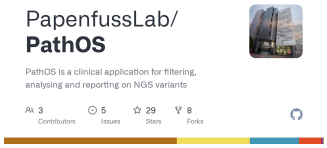

PathOS

A clinical decision support system for filtering, analyzing, curating, and reporting high-throughput sequencing (NGS) variants in a diagnostic laboratory setting.

https://github.com/PapenfussLab/PathOS

Overview

PathOS (Pathology Operating System) is an open-source, web-based clinical decision support system developed by the PapenfussLab at the Peter MacCallum Cancer Centre. Its primary function is to manage, analyze, and report on DNA sequencing variants from patient samples, translating raw data into clinically useful information.

Product Overview and Key Benefits

PathOS was developed to address the analysis and reporting bottlenecks in clinical high-throughput sequencing (NGS) workflows. It provides a robust, auditable laboratory workflow necessary for clinical diagnostics. While its genesis was in cancer molecular diagnostics, the system is broadly applicable to general NGS clinical reporting. The software is designed for reliable, consistent, and efficient reporting in a clinical laboratory setting.

Main Features and Capabilities

Variant Analysis and Filtering: PathOS identifies and filters out technical artifacts from sequencing data. It supports customizable and preset filter templates for different assays, including germline and somatic panels.

Variant Curation and Annotation: The system curates DNA changes, including single nucleotide variants (SNVs), insertions and deletions (indels), copy number variants (CNVs), and structural variants (SVs). It matches mutations with internal and external databases to identify known pathogenic or actionable mutations.

Clinical Reporting: PathOS renders the final, curated variants into a clinical diagnostic report suitable for the treating clinician, incorporating clinical evidence and relevant publications.

Workflow Management: It provides an auditable workflow for the entire analysis and reporting process, ensuring integrity and reproducibility.

Technology: The application is web-based, implemented in Java, Javascript, Groovy, and Grails, and uses MariaDB (MySQL compatible) for data storage. The bioinformatics pipeline is implemented using the Bpipe framework.

Target Users and Use Cases

Target Users: Clinical variant curators, clinical scientists, and pathology laboratories.
Use Cases: Routine clinical reporting of germline and somatic cancer samples, general NGS clinical reporting, and management of large-scale genomic research studies.

Key Features

- NGS Variant Filtering
- Clinical Variant Curation
- Clinical Diagnostic Report Generation
- Integration with LIMS/HL7
- Somatic and Germline Sample Reporting
- Actionable Mutation Identification
- Auditable Laboratory Workflow

Pricing

Model: free

PathOS is an open-source project available for free via source code and Docker images under the GNU General Public License v3.0. Deployment requires local infrastructure and technical expertise.

Target Company Size: medium, enterprise

Integrations

LIMS systems, Hospital Records Systems (via HL7), Global Variant Databases, Apache Lucene (Search Engine), Bpipe (Pipeline Framework)