NxClinical 4.2

One Assay. One Software. One Workflow.

A single system for analysis and interpretation of genomic aberrations from microarray and NGS.

SINGLE, PLATFORM-INDEPENDENT SYSTEM

Support for all major instrument vendors for both NGS & microarray

Simultaneous review of of sequence variants, AOH, and CNVs

Copy number estimation from NGS using the gold standard in CNV calling

CENTRALIZED STORAGE AND MANAGEMENT

Automatically builds a case history of all samples analyzed

Multi-user system with audit trail and global accessibility

Maintains data fidelity with a secure, central database

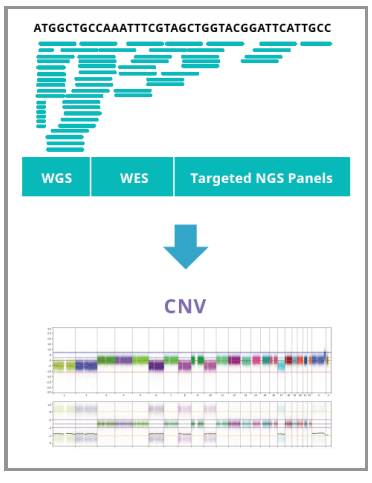
AUTOMATION & EXTENSIVE INTERPRETATION TOOLS

Customized workflows with adherence to ACMG or other guidelines

Single test paradigm – CNV, AOH, and sequence variants from NGS

Increased efficiencies across all areas saving time and money

N_XClinical 4.2 Benefits

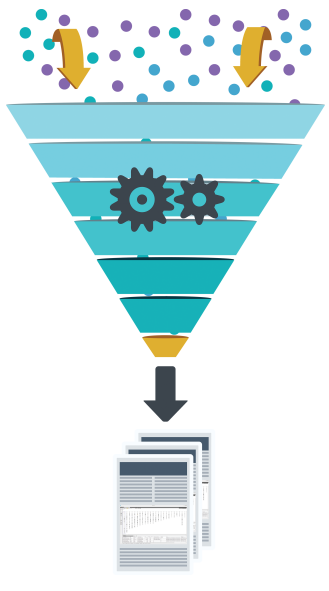


Interpret large and small genomic variants obtained from a single NGS test

- CNV estimation from NGS data (WES, WGS, and targeted NGS panels) using BioDiscovery's best in class algorithms (the gold standard in CNV calling)
- Replacement of multiple tests (CMA + NGS) with one, decreasing costs and time-to-diagnosis while increasing diagnostic yield
- Integration of array, NGS, and phenotypes in a single database with powerful querying by event, phenotype, sample attributes, and more
- Interactive visualization of raw data - probe plots, read depth plots, individual reads, single bases - for visual confirmation of events

Interactive filtering for rapid interpretation and reporting

- Apply virtual gene panels to limit review and interpretation to genes of interest for a specific test with easy step through inspection of each gene
- Filter based on event type, size, consequence, variant frequency, overlapping events, variant consequences, event severity, and more
- Perform trio analysis using sophisticated inheritance pattern filters
- Prioritize variants based on scoring of matches between clinical features and disease genes using HPO terms
- Pre-classify events based on prior classification of similar events in case history using an automated decision tree variant interpretation assistance system

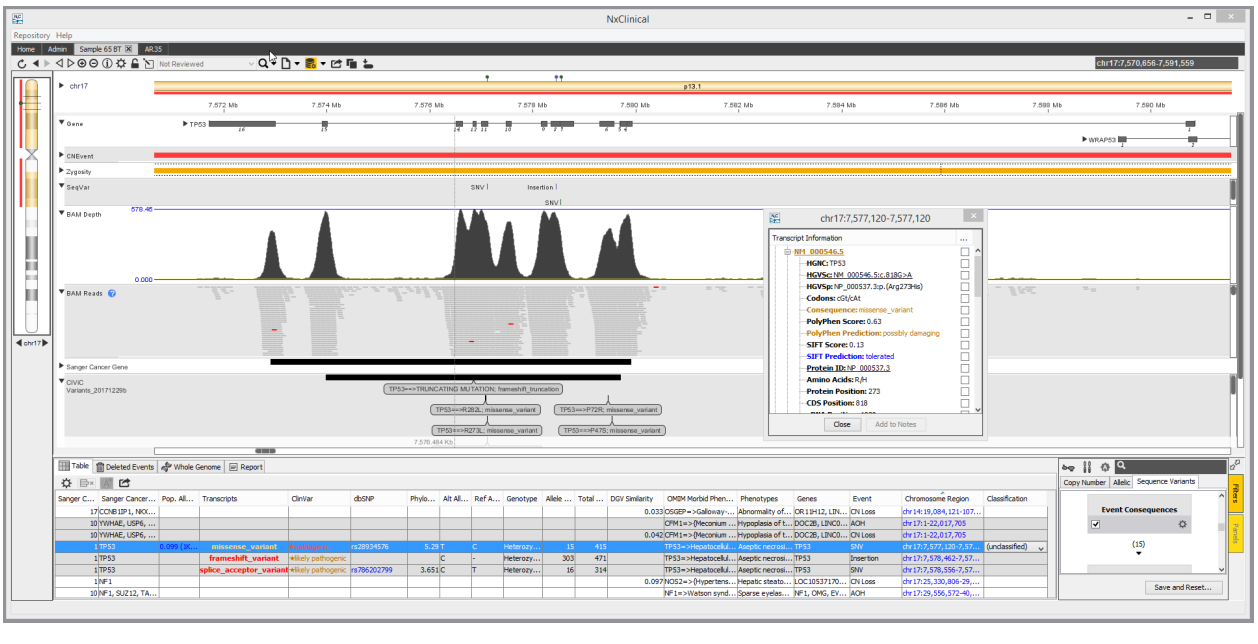


Real-time access to previous cases, annotations, and integrated reference databases

- Immediately view similar events in the lab case history and in public databases to events in the case under review
- Instantly access up-to-date integrated external databases (e.g. ClinGen/ClinVar, OMIM®*, DECIPHER, ExAC, CIViC) to aid with interpretation
- Maintain consistency across the enterprise with Admin controlled track updates and version locking upon case review completion

Shorter turnaround and increased consistency with extensive automation

- Decrease costs for deployment, maintenance, and training with a single system
- Automate processes to ensure consistency and compliance with regional guidelines (ACMG)
- Define sample types and workflows to quickly apply settings specific for each platform/test
- Maintain data integrity with access controls for multiple users and audit-trail for traceability



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