N_xClinical 4.1 Feature Highlights

N_xClinical supports the latest technologies and platforms

- Support for Thermo Fisher's newest platform CytoScan XON arrays for exon level CNV analysis.
- Support for Thermo Fisher's CYCHP file format (contains CNV and AOH calls) generated from their ChAS software or the command line programs, Affymetrix Power Tools (APT). This allows users who already have legacy data with events calls from these tools to load that data into N_xClinical rather than reprocessing the CEL files.

Enhancements to the filter chain for narrowing down potentially causative variants

- The recessive inheritance pattern filter shows compound heterozygous events overlapping the same gene and those with only a partial copy number gain event coinciding with the gene.
- The interest level filter can now be fined tuned to show individually selected consequences/interest levels rather than a range only.
- Population frequency, read depth, and quality of sequence variants no longer need to be selected during sample processing but now can be dynamically adjusted within the filter chain.

New features aiding sample review and interpretation

- Transcript annotations that were only available via a popup window can now be added as columns to the results table: e.g. HGVS nomenclature, functional effects from prediction algorithms, consequence, …
- Regulatory region annotations have been added to the results table as individual columns as well as in a pop up window similar to the transcript annotations.
- "Parent of Origin" field in the results table indicates the source of the affected allele for CNV and AOH events if at least one parent is linked to the proband.
- New region tracks such as CIViC, imprinted genes from Gene Imprint, and segmental duplications from UCSC are included with the installer. Many new focused files that can be used for the decision tree for pre-classification have been added: e.g. ClinGen Dosage Sensitive Map genes are divided into several regions such as Triplosensitivity Pathogenic, Triplosensitivity Benign, Haploinsufficiency Pathogenic… and DECIPHER Development Disorder Genotype – Phenotype Database (DDG2P) is separated as Dominant, Recessive…
- Changes to the html report layout improve readability as well as displays richer annotations.

Improvements to Sample Types and Processing

- Improvements in processing include the ability to define different processing servers for different Sample Types allowing WGS samples for example to be run on a more powerful server than smaller array files which can be run on a less powerful server.
- User can better customize Sample Attributes to create their own family relationship labels to best represent their workflow or the local language.
- Multiple siblings can be linked to a single sample with the affected status separated from the sample label.