Illumina Connected Annotations v3.23.0 Software Release Notes

INTRODUCTION

These Release Notes detail the latest release of Illumina Connected Annotations, previously known as Nirvana, including known issues.

Connected Annotations provides clinical-grade annotation of genomic variants (SNVs, MNVs, insertions, deletions, indels, STRs, gene fusions, and SVs (including CNVs)).

NEW FEATURES

- Encrypted data format for paid supplementary annotation data sources
- Pass ID field from VCF file to JSON
- Added PhyloPPrimate to PhyloPPrimate supplementary annotation data sources
- Implement license feature for paid supplementary data sources

UPDATED FEATURES

- Added qualitative classifications for PrimateAI-3D
- Provided tiers for COSMIC Cancer Gene Census

DATA UPDATES

- Updated ClinGen DSM & GDV, OMIM, and ClinVar
- Updated COSMIC to v99

RESOLVED ISSUES

- Omim downloader version path fix
- Nirvana failing to annotate on alt contigs due to missing contig sequence in reference
- Annotator always exits with 0 even with exception
- Downloader in Prod uses manifest file from Dev

UNRESOLVED ISSUES

- Nirvana incorrectly determines gnomAD low complexity regions at translocation breakend boundaries
- Nirvana incorrectly handles insertions where RNA-edit insertions already occur

KNOWN LIMITATIONS

Issue ID	Summary	Remedy/Workaround
NIR-919	Nirvana does not handle protein sequences that are partially aligned to the genome	No workaround. Fix planned for future version
NIR-918	Nirvana does not handle ribosomal frameshifts properly during translation	No workaround. Fix planned for future version
NIR-917	Nirvana does not truncate deletions that start within a deletion RNA-edit (HGVS c.)	No workaround. Fix planned for future version
NIR-916	Nirvana does not detect HGVS c. duplications when inserting a base into a deletion RNA-edit	No workaround. Fix planned for future version
NIR-915	Nirvana does not convert SNVs that land in a deletion RNA-edit as an insertion (HGVS c.)	No workaround. Fix planned for future version

RELEASE HISTORY

Revision	Release Reference	Originator	Description of Change
00	1105124	Rajat Roy	Initial release