Illumina Connected Annotations v3.22.0 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

- Add MANE select tag to transcripts
- Consequence Impact

UPDATED FEATURES

- PrimateAI-3D
- Release Cache 32
- Update VF calculation on MT to reflect changes in DRAGEN variant calling
- DRAGEN Somatic Caller MT VF Fix

DATA UPDATES

- October 2023 data update (ClinGen DSM & GDV, OMIM, ClinVar, and dbSNP)

RESOLVED ISSUES

- Nirvana crashing at HGVS p. IsStartLost
- NirvanaLambda failing to load Tabix index
- Variant Rotator: prevent right shift for variants not in region
- Nirvana Lambda: Tabix indexing is not moving the cursor to the beginning of the VCF line
- Nirvana lambda returns a null in files list
- Nirvana should simplify HGVS p. notation for deletions that disrupt the initiation codon
- SingleAnnotation behaves differently in Lambda vs REST API

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	1095450	Rajat Roy	Initial release