

Human Whole Genome Sequencing Service

Informatics Pipeline Release Notes

Version 4.0.2

1. Internal workflow changes to automatic quality control outputs.
2. Stability updates.

Version 4.0.0

NEW FEATURES:

3. Upgraded Isaac to version iSAAC-SAAC00776.15.01.27
4. Isaac
 - Additional alignment attempted on reads. Now comparable to bwa-mem
5. Upgraded SV calling algorithm to Manta manta-0.23.1.
6. Upgraded CNV calling algorithm to Canvas 1.1.0.5.
7. Updated IsaacVariantCaller to 2.1.4.2
8. Updated the annotation pipeline and annotation sources.

Version 3.0.0

1. Upgraded Isaac to version iSAAC-01.14.02.06
2. Removed GATK indel realignment.
 - Now performed by Isaac inline.
3. Upgraded SV calling algorithm from Grouper to Manta manta-0.18.1.
4. Upgraded CNV calling algorithm from CNVseg to Canvas 1.1.0.1.
5. Changes to PDF report.
 - Now counts all alternative alleles as separate events.
 - Removed outer SNV gene name track from circos plot.
 - SV counts, in the absence of another calculated value, defaults to 0 counts.
 - Only counts passing statistics.
6. Changed annotation.
 - Now using RefSeq as annotation base.
 - Upgraded to Ensembl version 72.
 - Dropped HGMD annotation.
 - Added ClinVar annotation.

Version 2.0.2

1. Internal pipeline changes for improved stability and quality control. No changes to output.

Version 2.0.11

1. Modifications to the PDF report.
 - Auto scale the coverage plot for deep sequencing projects.
2. Modification to annotation pipe to:
 - Remove source entries.
 - Alter order of vep CSQ filtering for tri-allelic corner cases.
3. Updated to VEP 2.7.

Version 2.0.1

1. Workflow change to include reporting of non-pass variants for SNPs and indels.
2. Minor graphical changes to summaryreport.pdf output.

Version 2.0.0

1. Initial Post-Casava release.
2. Changed Aligner from Eland to Isaac iSAAC-01.13.04.29.
3. Significant changes to VCF file structure.
4. Reworked PASS/FAIL VCF filtering parameters.
5. Added PDF report.