

WiGiTs Oncoanalyser v2.3 Release Notes

Overview

The v2.3 pipeline release has these key improvements:

- Improvements to Sage germline filters
- Copy number visualisations and output files
- Cider performance improvements
- Heterozygous deletions for panels

Resource File Updates

Tool / Stage	Description
Driver gene panel	Added het deletion and amplification ratio
Sage	Add hotspots for ASXL1, TCF7L2 and BAX, remove ATR hotspot Removed ASXL1 from v37 PON
Purple	Germline deletion cohort frequency file following hg38 re-run

Tool Updates

See tool releases and READ-ME for full details.

Tool	Version	Release Notes and Key Changes
BamTools	1.5	Handle unpaired reads Include fully-unmapped reads in summary metrics Awareness of dropped duplicates when enabled in Redux
Cider	1.1	Use BWA instead of BlastN for alignment
Cobalt	2.2	Ignore IG pseudogene regions
Esvee	1.2	Minor fix for depth annotation in unmapped regions
Lilac	1.7.2	Solution complexity calculation: Split long exons in half Limit exon boundary filtering to 10+ fragments overlapping an exon boundary Add B*40:278 to allele blacklist
Linx	2.2	Germline DEL-DUP disruptions remain reportable if in 3'UTR Visualiser: plot HET deletions and added new CN events plot
Pave	1.8.1	Set v37 PON filters automatically rather than assuming v38
Purple	4.3	Targeted panel region copy number output file Chromosome arm copy number output file Report heterozygous deletions Fix germline homozygous deletion bug Recovery of germline SV if TINC identified
Sage	4.2	Improve handling of long homopolymer hotspots Improved germline filters Don't count CORE support to AD/AF if exceeds 25% of total support

Configuration Changes

Tool	Release Notes and Key Changes
BamTools	BamMetrics writes target region file in panel mode
Redux	Write duplicate statistics
Sage	Enable calling MT variants
Purple	Driver amplification ratio and het deletion thresholds can be set in driver gene panel

Technical Changes

Tool	Performance Characteristics
Cider	Switch to BWA from BlastN gives 10x alignment performance improvement