

WiGiTs Oncoanalyser v2.2 Release Notes

Overview

The v2.2 pipeline release has these key improvements

- Esvee improved artefact detection
- Targeted panel resource file generation routine
- Longitudinal pipeline for ctDNA and other samples
- Improved copy number fitting and deletion calling for targeted panels
- Improved HLA typing for targeted panels
- TINC detection in Sage

Resource File Updates

Tool / Stage	Description
Pave	Removed 58 variants from hg19 WGS PON and 57 variants from hg38 WGS PON
Known fusions	UBTF exon DEL-DUP, TERT enhancer downstream region added
Driver gene panel	Removed SPATA31A7
Sage	No longer uses actionable and gene coverage files, derived from driver gene panel PON regenerated for hg38
Esvee	No longer uses blacklist file PON regenerated using v1.1
Lilac	IPD-IGMT/HLA database (allele sequences) - 2024-12-19 Allele Frequency Net Database (AFND) - 2025-01-28
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
Amber	4.2	Lower min VAF threshold Raise <u>min_base_qual</u> from 13 to 30
<u>BamTools</u>	1.4	Gene coverage & exon mean coverage moved from Sage to <u>BamMetrics</u>
Cider	1.0.4	
Cobalt	2.1	Use realised GC content, widen range (0.24 - 0.68), smooth points Only use on target regions for GC <u>normalisaiton</u>
<u>Esvee</u>	1.1	Various changes to remove artefacts Fixed issues relating to read assignment and assembly extension Improved homology identification Ignore SVs outside panel regions Improved PON matching
Lilac	1.7	Allele frequencies now based on global population frequencies from ANFD Replace wildcards in IMGT/HLA allele sequences with consensus 2-digit allele sequences Improved handling of variable coverage Filter nucleotide/amino acid candidates based on relative (rather than absolute)