

GENIE 12.0-public release notes

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July 12, 2022

Release notes

- 19208 samples added
- **CHOP**
 - Fusion panel includes genes that are not on the other panels/pipelines that generate the SNV and indel calls. No BED file for fusion panel.
 - Confirmed genomic data for **CHOP-STNGS** samples contain no artifacts.
- **CRUK**
 - Confirmed genomic data for **CRUK-TS** samples contain no artifacts.
- **DFCI**
 - Expected discrepancies in expected and submitted gene count because of regions that are targeted for calling structural rearrangements.
- **UCSF**
 - Confirmed genomic data for **UCSF-NIMV4** samples contain no artifacts.
- **UHN**
 - Confirmed genomic data for **UHN-54-V1** samples contain no artifacts.
 - Cannot guarantee that there are no artifacts for the genomic data of **UHN-555** samples, but have done the best in removing most.
- **YALE**
 - Intentionally reports only amplifications in copy number data due to internal policy.
 - **YALE-OCF-V2** panel's expected gene count is 134 because one intron is included. Gene panels are created with only exons hence the difference in actual vs expected gene counts.

Data Concerns/Issues

These are the known data issues for this release. **Note:** There could be more undiscovered issues.

- Foundation Medicine genomic regions discrepancy
 - **DUKE/WAKE** use the Foundation Medicine T5A, T7, DX1, R2D2 bait sets, but some of the bed files uploaded don't seem to match the expected gene count per panel.
- Sites/panels with mismatching information for the number of genes in `assay_information.txt` vs `genomic_information.txt`.
 - **GRCC, SCI, UCHI, WAKE,**
- Not all variants have variant counts (t_depth, t_alt_count, t_ref_count)
- Genome Nexus related issues
 - Duplicated variants
 - Non-somatic mutations (Reference_Allele == Tumor_Seq_Allele1 == Tumor_Seq_Allele2)
 - SNV variants annotated as DNP or ONP