GENIE 19.0-public release notes

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Release notes

These release notes will encompass the changes made in the 19 consortium series and general center information.

- 21819 samples have been added since the last public release.
- New panel(s) since the last public release:
  - VICC-02-XFV2
  - UHN-WGS-V1
  - UHN-MEL-7
  - UHN-MEL-12
  - UHN-MEL-11
  - UHN-CST-8
  - UHN-MEL-13
  - UHN-MEL-34
  - UHN-CST-11
  - UHN-CST-7
  - UHN-HP-8
  - UHN-HP-2
  - UHN-HM-26
  - UHN-HM-29
  - UHN-CST-3
  - UHN-HP-1
  - UHN-CST-43
  - UHN-HPV3-19
  - UHN-CST-2
  - UHN-HPV4-19
  - UCHI-ONCOPLUS119-V1
  - UCHI-ONCOPLUS147-V1
  - UCHI-ONCOPLUS154-V1
  - UMIAMI-FD-F1CDX
  - UMIAMI-FD-F1H
  - UMIAMI-FD-F1L
  - UMIAMI-GD-360
  - CHOP-HEMEP-V2
  - GRCC-2022569
  - GRCC-2022572
  - GRCC-2106321
  - GRCC-2112732
  - VICC-02-XFV3
  - YALE-HSM-V1
  - YALE-OCP-V2
- 17.0-package released. See all updates here.
  - Upgraded pandas, synapseclient, and supported Python versions.

  - Pediatric unmasking
- Genome-Nexus Update: Previously, this annotator used newer gene symbol aliases when annotating variants. In 19.4, this issue has been resolved — annotations now use the gene symbols defined in GRCh37.

Data Concerns/Issues

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

- The cluster migration–related annotation issue identified in the 19.1-consortium release has been resolved in 19.2
- PROV's low variants issue has been resolved in 19.3
- UCHI's maf data for 19.4 may contain newer gene symbols (e.g. H3-3A instead of H3F3A) due to a processing issue
- In the sample file, there are two JHU patients with AGE_AT_SEQ_REPORT and AGE_AT_SEQ_REPORT_DAYS values that are negative.
- The unexpectedly large amount of failed annotations for PROV and UMIAMI have been resolved in 19.5
- The number of expected genes in certain gene panels may be incorrect due to sites submitting invalid gene symbols, coordinates, or incorrect assay information.
- **CRUK**
  - Confirmed genomic data for **CRUK-TS** samples contain no artifacts.
- **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.
- **MSK**
  - Confirmed genomic data for MSK HEME panel samples contain no artifacts.
- **DFCI**
  - Expected discrepancies in expected and submitted gene count because of regions that are targeted for calling structural rearrangements.
- **UHN**
  - Confirmed genomic data for **UHN-54-V1** samples contain no artifacts.
  - UHN has some duplicate bed regions in their processed data. This is a processing issue, no action is needed from sites.
- **YALE**
  - Intentionally reports only amplifications in copy number data due to internal policy.
  - **YALE-OCP-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.
- 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.
- Not all variants have variant counts (t_depth, t_alt_count, t_ref_count).
- Duplicated variants listed from **VICC** are expected due to providing Tempus samples.
- Genome Nexus related issues:
  - Duplicated variants