GENIE 16.0-public release notes

Sage Bionetworks

June 26, 2024

Release notes

These release notes will encompass the changes made in the 16 consortium series releases and general center information.

- 16507 samples have been added since the last public release.

Data Concerns/Issues

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

- 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.
- **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
  - Non-somatic mutations (Reference_Allele == Tumor_Seq_Allele1 == Tumor_Seq_Allele2)
  - SNV variants annotated as DNP or ONP

Genome-Nexus version

- **genomeNexus**
  - **server**

        * Version: “1.0.2”
        * Static: true
    - **database**
        * Version: “3.6.2”
        * Static: true
- **vep**
    - **server**
        * Version: “NA”
        * Static: true
    - **cache**
        * Version: “NA”
        * Static: true
    - Comment: null
- **annotationSourcesInfo**
    - **VEP**
        * Version: “grch37”
        * Type: “mirrored”
        * Description: “VEP determines the effect of your variants (SNPs, insertions, deletions, CNVs or structural variants) on genes, transcripts, and protein sequence, as well as regulatory regions.”
        * URL: “https://grch37.ensembl.org/info/docs/tools/vep/index.html”
    - **Cancer Hotspots**
        * Version: “v2”
        * Type: “mirrored”
        * Description: “A resource for statistically significant mutations in cancer”
        * URL: “https://www.cancerhotspots.org”
    - **3D Hotspots**
        * Version: “v2”
        * Type: “mirrored”
        * Description: “A resource for statistically significant mutations clustering in 3d protein structures in cancer”
        * URL: “https://www.3dhotspots.org/”
    - **HGNC**
        * Version: “22-10-01”
        * Type: “mirrored”
        * Description: “The resource for approved human gene nomenclature. Genome Nexus uses HGNC gene symbols in annotation”
        * URL: “http://ftp.ebi.ac.uk/pub/databases/genenames/hgnc/archive/monthly/tsv/”
    - **reVUE**
        * Version: “d8a7d01bac02671e74b4522bacfca6e82f360046”
        * Type: “mirrored”
        * Description: “A Repository for Variants with Unexpected Effects (VUE) in Cancer”
        * URL: “https://www.cancerrevue.org/”
    - **My Variant Info**
        * Version: “Includes many annotation sources, see https://docs.myvariant.info/en/latest/doc/data.html”
        * Type: “external”
        * Description: “MyVariant.info provides simple-to-use REST web services to query/retrieve variant annotation data, aggregated from many popular data resources.”
        * URL: “https://myvariant.info”
    - **Mutation Assessor**
        * Version: “v3”
        * Type: “mirrored”
        * Description: “Mutation Assessor predicts the functional impact of amino-acid substitutions in proteins, such as mutatio. . . ”