applied biosystems

CarrierScan™ Reporter 1.0 Release Notes

(For research use only. Not for use in diagnostic procedures.)

CarrierScan Reporter 1.0 includes the following features:

1. Controlled sample carrier state determinations
2. Confidence of carrier state assessments
3. Individual residual chance of carrier occurrence
4. Paired residual chance of phenotype occurrence
5. Compiled information from public databases for associated phenotypes
6. Customizable reports

Software and Hardware Requirements:

CarrierScan Reporter 1.0 is only supported on 64-bit systems with the following operating systems:

- Windows 7 Professional SP1 (64-bit)
- Windows 10 Professional (64-bit)

Memory and CPU Recommended Requirements

- Quad Core System, 2.83 GHz or above
- Minimum 8GB RAM, recommended 16GB RAM

The following are the known issues that exist in CarrierScan Reporter 1.0:

1. (CS-301) If a search phrase is on a table only once, the first time it is searched for, it will be found and highlighted. Subsequent searches will result in “the string isn’t found on the table” and the original row it was found in will remain highlighted.
2. (CS-476) On Windows 10 operating system, the checkbox display looks like cutoff edit boxes. It is a display issue but does not affect the use of the software.
3. (CS-534) When sorting on the “Mutations Detected” column, the application is not able to sort any column that contains both numeric and alpha characters in the name. All tables in the software are recorded in .txt files that can be opened in spreadsheet applications such as Microsoft Excel to support alphanumeric sorting needs.
4. (CS-563) When using a proxy server, there is a short time delay between entering a valid proxy user/password to the verification process.
5. (CS-579) If the “Create CN Visualization Files” option is selected, there will be no warning message if there are missing *cnv.txt files for samples in the Axiom Analysis Suite results folder. The software will only create visualization data for samples with existing *cnv.txt files. If there are missing samples, please reanalyze the files in Axiom Analysis Suite and reload the new results into CarrierScan Reporter.
6. (CS-760) Mutations on Chromosome X for Male samples will be shown as haploid when (AA/BB/CC/DD/EE) are represented if het (AB/AC/AD/AE/BC/BD/BE/CD/CE/DE) the appropriate diploid call will be translated.
7. (CS-796) The analysis name sorting is based upon Microsoft Windows sorting, which sorts 1 character at a time. The sorting is 1, 19, 9 instead of 1, 9, 19.
8. (CS-1048) Analysis results cannot be refreshed/updated if a report or file is opened.

9. (CS-1059) On occasion, CarrierScan viewer may stop working. This is a low occurrence. If this occurs, reboot the machine.
10. (CS-1091) In order to have CN samples display, be sure that during an analysis, no files are in use or open. If they are open, please close it, otherwise information will not be updated during the analysis run.
11. (CS-1141) Paired Residual Chance of Occurrence calculations require that all 3 columns for the assigned ethnicities to be present. When any number of these columns are missing an error prompt will attempt to describe the missing columns, but sometimes it may only correctly identify some of the missing columns. Please review the panel to ensure all 3 columns as described in the user guide exist for all assigned ethnicities.
12. CarrierScan Reporter is designed to work with Axiom Analysis Suite 'Best Practices Workflow' analysis of CarrierScan arrays. If using APT instead of Axiom Analysis Suite to analyze raw data (CEL files), then the results should be organized in the same folder structure to be readable into CarrierScan Reporter. Please follow the APT best practices workflow as defined for CarrierScan analysis to generate the necessary files in the appropriate format and organization.
13. When analyzing Sample Carrier Status for variants on HBA1 and HBA2, the software will consider these genes together as if they were the same gene to support Carrier assessment across both genes according to NCBI guidelines (https://www.ncbi.nlm.nih.gov/books/NBK1435).