Copy Number Variant Annotation Pipeline File Format Specification Sheet

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Revision History

Revision	Notes
A	Initial document release

Introduction

The Variant Annotation Pipeline enables users to determine if a Bionano copy number variant (CNV) call is relevant to certain phenotypes or disease traits. For more information about the pipeline, please refer to Bionano Solve Theory of Operation: Variant Annotation Pipeline (PN 30190). The output file of the Variant Annotation Pipeline is an annotated CNV results file, with additional annotation columns appended. The CNV file format is a general format to describe CNVs detected by Bionano; please refer to Bionano Solve Theory of Operation: Structural Variant Calling (PN 30110) for details on the CNV calling algorithm and output files.

This document describes only the additional annotation columns. Also, note that the last few columns can vary depending on whether a trio, dual or single analysis has been performed upon execution of the Variant Annotation Pipeline.

Annotation Columns – All Analyses

Statistic	Description
OverlapGenes	A semi-colon separated list indicating which genes overlap with the CNV.
NearestNonOverlapGene	The next closest gene to the CNV.
NearestNonOverlapGeneDistance	The distance between the CNV and the next closest
num_overlap_DGV_calls	If the sample is human, then the CNVs would be compared against the Database of Genomic Variants (DGV), and the number of DGV variants overlapping the call is outputted.
UCSC_web_link1	If the sample is either human or mouse, then a weblink to the CNV region in the UCSC genome browser would be created.
ISCN	If the sample is human, then the CNVs would be annotated with the International System for Human Cytogenomic Nomenclature (ISCN) notation for CNV

Trio Analysis

Statistic	Description
Found_in_parents	Whether the CNV call is also identified in the father's or mother's assembly. The possible values are 'mother', 'father', 'both' and 'none'.

Dual Analysis

Statistic	Description
Found_in_control_paired	Whether the CNV call is also identified in the control sample's assembly. The possible values are 'yes' or 'no'.

Technical Assistance

For technical assistance, contact Bionano Genomics Technical Support.

You can retrieve documentation on Bionano products, SDS's, certificates of analysis, frequently asked questions, and other related documents from the Support website or by request through e-mail and telephone.

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