



# **Copy Number Variant Annotation Pipeline File Format Specification Sheet**

Document Number: 30461

Document Revision: A

## Table of Contents

|                            |   |
|----------------------------|---|
| Legal Notice .....         | 3 |
| Introduction .....         | 4 |
| Trio Analysis .....        | 5 |
| Dual Analysis .....        | 6 |
| Technical Assistance ..... | 7 |

## Legal Notice

---

### **For Research Use Only. Not for use in diagnostic procedures.**

This material is protected by United States Copyright Law and International Treaties. Unauthorized use of this material is prohibited. No part of the publication may be copied, reproduced, distributed, translated, reverse-engineered or transmitted in any form or by any media, or by any means, whether now known or unknown, without the express prior permission in writing from Bionano Genomics. Copying, under the law, includes translating into another language or format. The technical data contained herein is intended for ultimate destinations permitted by U.S. law. Diversion contrary to U. S. law prohibited. This publication represents the latest information available at the time of release. Due to continuous efforts to improve the product, technical changes may occur that are not reflected in this document. Bionano Genomics reserves the right to make changes in specifications and other information contained in this publication at any time and without prior notice. Please contact Bionano Genomics Customer Support for the latest information.

BIONANO GENOMICS DISCLAIMS ALL WARRANTIES WITH RESPECT TO THIS DOCUMENT, EXPRESSED OR IMPLIED, INCLUDING BUT NOT LIMITED TO THOSE OF MERCHANTABILITY OR FITNESS FOR A PARTICULAR PURPOSE. TO THE FULLEST EXTENT ALLOWED BY LAW, IN NO EVENT SHALL BIONANO GENOMICS BE LIABLE, WHETHER IN CONTRACT, TORT, WARRANTY, OR UNDER ANY STATUTE OR ON ANY OTHER BASIS FOR SPECIAL, INCIDENTAL, INDIRECT, PUNITIVE, MULTIPLE OR CONSEQUENTIAL DAMAGES IN CONNECTION WITH OR ARISING FROM THIS DOCUMENT, INCLUDING BUT NOT LIMITED TO THE USE THEREOF, WHETHER OR NOT FORESEEABLE AND WHETHER OR NOT BIONANO GENOMICS IS ADVISED OF THE POSSIBILITY OF SUCH DAMAGES.

### **Patents**

Products of Bionano Genomics® may be covered by one or more U.S. or foreign patents.

### **Trademarks**

The Bionano Genomics logo and names of Bionano Genomics products or services are registered trademarks or trademarks owned by Bionano Genomics in the United States and certain other countries.

Bionano Genomics®, Saphyr®, Saphyr Chip®, and Bionano Access® are trademarks of Bionano Genomics, Inc. All other trademarks are the sole property of their respective owners.

No license to use any trademarks of Bionano Genomics is given or implied. Users are not permitted to use these trademarks without the prior written consent of Bionano Genomics. The use of these trademarks or any other materials, except as permitted herein, is expressly prohibited and may be in violation of federal or other applicable laws.

© Copyright 2021 Bionano Genomics, Inc. All rights reserved.

## 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   |
|--------------------------------------|---|
| <b>OverlapGenes</b>                  | A semi-colon separated list indicating which genes overlap with the CNV.  |
| <b>NearestNonOverlapGene</b>         | The next closest gene to the CNV.   |
| <b>NearestNonOverlapGeneDistance</b> | The distance between the CNV and the next closest   |
| <b>num_overlap_DGV_calls</b>         | 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. |
| <b>UCSC_web_link1</b>                | If the sample is either human or mouse, then a weblink to the CNV region in the UCSC genome browser would be created.   |
| <b>ISCN</b>                          | 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  |
|-------------------------|--|
| <b>Found_in_parents</b> | 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  |
|--------------------------------|--|
| <b>Found_in_control_paired</b> | 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.

| Type    | Contact   |
|---------|---|
| Email   | <b>support@bionanogenomics.com</b>  |
| Phone   | <b>Hours of Operation:</b><br><br><b>Monday through Friday, 9:00 a.m. to 5:00 p.m., PST</b><br><br><b>US: +1 (858) 888-7600</b> |
| Website | <b><a href="http://www.bionanogenomics.com/support">www.bionanogenomics.com/support</a></b>                                     |