Global Screening Array Manifest File Release Notes

v1.0 C1

May 17, 2018

FOR RESEARCH USE ONLY

Introduction

These release notes detail the key changes to the Global Screening Array manifest files (.bpm &.csv formats) for v1.0 C1 since the v1.0 A1 initial release.

NEW FEATURES:

- None

IMPROVEMENTS:

- Optimized for CNV detection
- Validated correlation against 1000 Genomes data
- Assigned annotations for MapInfo 0 markers
- Updated annotations for multinucleotide variants
- Removed rsIDs with MapInfo discrepant against dbSNP 150

ADDITIONAL SUPPORT FILES:

- List of markers removed from manifest A1 and reason(s) for removal
    - Increased Correlation Stringency: Validated correlation against 1000 Genomes data
    - Discrepant rsID: MapInfo discrepant against dbSNP 150
    - Multimapper: Markers that do not map or do not have a unique location in GRCh37
    - Poor Clustering: Undefined or nondistinct clusters
- List of multinucleotide variants
- VCF file for multinucleotide variants
- List of tri- and tetrallelic markers*
- Updated cluster file

* Illumina does not officially support the design of assays for triallelic SNPs and multinucleotide variants, as the Infinium technology is specifically intended for assaying only biallelic SNPs. These designs are included in the Global Screening Array at the request of the designing consortium. Because we do not support this type of design, we cannot assist in the interpretation of such assays.

Revision History

Revision	Date	Description of Change
v1.0 C1	17-May-18	Validated for correlation against 1000 Genomes data and dbSNP annotations
v1.0 A6	8-Jul-17	Optimized for CNV detection
v1.0 A1	10-Jan-17	Initial release