BlueFuse Multi Color Key

CytoChip Module

DecisionTrack Key

Track	Color	Genome Build Availability
Current	Regions of copy number change in the current sample.	GRCh37 GRCh38
	Duplication or gain of material	
	Deletion or loss of material	
BAC Gain/Loss	All CNV regions compiled from the BAC array experiments in the current database are presented as a compressed track.	GRCh37
	Present in > 50% of experiments in the database.	
	Present in 10–50% of experiments in the database.	
	Present in 2–10% of experiments in the database.	
	Present in < 2% of experiments in the database.	
Oligo Gain/Loss	All CNV regions compiled from the oligo array experiments in the current database are presented as a compressed track.	GRCh37
	Present in > 50% of experiments in the database.	
	Present in 10–50% of experiments in the database.	
	Present in 2–10% of experiments in the database.	
	Present in < 2% of experiments in the database.	
Bead Gain/Loss	All CNV regions compiled from the BeadArray experiments in the current database are presented as a compressed track.	GRCh37 GRCh38
	Present in > 50% of experiments in the database.	
	Present in 10–50% of experiments in the database.	
	Present in 2–10% of experiments in the database.	
	Present in < 2% of experiments in the database.	

Track	Color	Genome Build Availability
Seq Gain/Loss	All CNV regions compiled from the sequencing-based experiments in the current database are presented as a compressed track.	GRCh37
	Present in > 50% of experiments in the database.	
	Present in 10–50% of experiments in the database.	
	Present in 2–10% of experiments in the database.	
	Present in < 2% of experiments in the database.	
Current LOH	The regions of loss of heterozygosity in the current sample.	GRCh37 GRCh38
SNP LOH	All regions of loss of heterozygosity from oligo SNP array experiments in the current database are presented as a compressed track.	GRCh37
	Present in > 50% of experiments in the database.	
	Present in 10–50% of experiments in the database.	
	Present in 2–10% of experiments in the database.	
	Present in < 2% of experiments in the database.	
Bead LOH	All regions of loss of heterozygosity from BeadArray experiments in the current database are presented as a compressed track.	GRCh37 GRCh38
	Present in > 50% of experiments in the database.	
	Present in 10–50% of experiments in the database.	
	Present in 2–10% of experiments in the database.	
	Present in < 2% of experiments in the database.	
Disease	Regions associated with constitutional disorders, targeted by additional probes in CytoChip array designs.	GRCh37
	Default color for disease regions.	
Genes	The set of Ensembl genes. This track can be shown as compressed or expanded. Additional links are provided to the genomic region in Ensembl[1], UCSC[2], DGV[3], OMIM[4], and HGNC[5].	GRCh37 GRCh38
	Gene has an OMIM disease annotation.	
	Default color for genes.	
Exons	Exons that are part of the canonical transcript of each gene from Ensembl.	GRCh37 GRCh38
	Protein-coding exons in the canonical transcript.	
	UTR exons or noncoding transcript.	

Track	Color	Genome Build Availability
Mask	This track is shown if a genome mask has been specified for the current sample.	GRCh37 GRCh38
	Areas excluded by an exclusion mask.	
	Areas included by an inclusion mask.	
DGV Gain/Loss	CNV data compiled from multiple studies of normal populations from the Database of Genomic Variants[3].	GRCh37 GRCh38
	Frequency is > 20% and evidence is from 3 or more studies.	
	Frequency is 10–20% and evidence is from 3 or more studies.	
	Frequency is 2–10% and evidence is from 2 or more studies.	
	All remaining regions with low frequency or high frequency CNVs that are only found in individual studies.	
DECIPHER Gain/Loss	Affected CNV data compiled from the DECIPHER project[6] and color coded according to classification. The tooltips for these tracks show summary phenotype information from the DECIPHER database. In the compressed track, phenotypes with a greater frequency than expected by chance are marked (eg, **NEUROLOGY**).	GRCh37 GRCh38
	CNV	
	No majority category (compressed tracks only)	
	Unknown/Unclassified	
	Familial	
	De Novo	
ISCA Gain/Loss	CNV regions potentially involved in intellectual and developmental disabilities. ISCA Consortium[7] members submit this data to dbVar.	GRCh37
	Benign	
	Pathogenic	
	No majority category (compressed tracks), or Unknown (expanded tracks)	
ISCA C Gain/Loss	Consensus (reviewed) regions involved in intellectual and development disabilities. The data are obtained from the ISCA Consortium[7] database.	GRCh37
	Benign	
	Pathogenic	
	No majority category (compressed tracks), or Unknown (expanded tracks)	

Track	Color	Genome Build Availability
dbVar	Human CNV regions with a clinical assertion from the dbVar database of genomic variants. Includes ISCA submissions.	GRCh38
	Benign	
	Pathogenic	
	No majority category (compressed tracks), or Unknown (expanded tracks)	

Infinium Control Data Key

Chart	Color	Data Set
Staining Red		Staining Red (High)
		Staining Red (Bgnd)
Staining Green		Staining Green (High)
		Staining Green (Bgnd)
Extension Red		Extension (A)
		Extension (T)
		Extension (C)
		Extension (G)
Extension Green		Extension (A)
		Extension (T)
		Extension (C)
		Extension (G)
Target Removal		Target Removal
Hybridization		Hyb (High)
		Hyb (Medium)
		Hyb (Low)
Stringency		String PM
		String MM
Nonspecific Binding Red		NSB (Bgnd 1)
		NSB (Bgnd 2)
		NSB (Bgnd 3)
		NSB (Bgnd 4)
Nonspecific Binding Green		NSB (Bgnd 1)
		NSB (Bgnd 2)

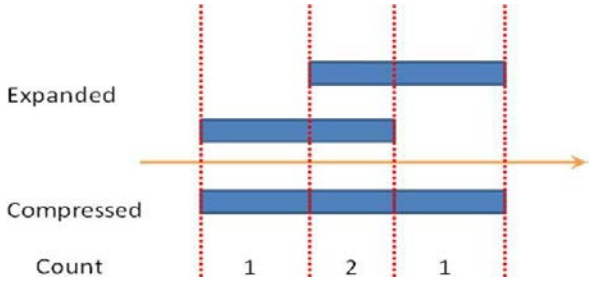
		NSB (Bgnd 3)
		NSB (Bgnd 4)
Nonpolymorphic Red		NP (A)
		NP (T)
		NP (C)
		NP (G)
Nonpolymorphic Green		NP (A)
		NP (T)
		NP (C)
		NP (G)
Restoration Red		Restore
Restoration Green		Restore

Compressed Tracks

Compressed tracks compress large data sets into a single track row. To compress tracks, BlueFuse Multi performs the following steps:

1. The overlapping regions are resolved into nonoverlapping segments.
2. The number of regions that contribute to each segment is counted.
3. Region labels are collected.

In this image, 2 overlapping regions are resolved into 3 segments that cover the original regions without overlapping.



The following bullets contain additional information about compressed tracks.

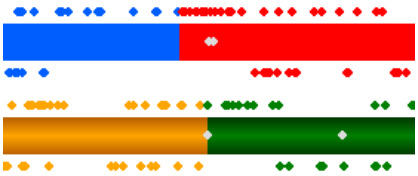
- Tracks that display CNVs from a population:
  - Colored according to CNV frequency.
  - Can be interpreted as a histogram.
  - Red and orange colors indicate regions of high frequency copy number variation.
- Tracks where the color represents phenotype information:
  - Colors of segments reflect the majority color of the corresponding regions in the expanded track.
  - Example: Emory tracks contain overlapping CNV regions, which are labeled as benign, pathogenic, or uncertain. In the compressed track, if most corresponding regions are marked as pathogenic, a segment is colored green (pathogenic).

Karyomapping Module

Trio (Paternal, Maternal, Reference) Chromosomes

Paternal Chromosome		Paternal chromosomes are shown in blue (P1) and red (P2).
Maternal Chromosomes		Maternal chromosomes are shown in orange (M1) and green (M2)
Reference Chromosomes		The reference chromosome pair is arbitrarily assigned as P1 (blue) and M1 (orange).

Embryo Chromosomes

Embryo Chromosomes 	The paternally inherited embryo chromosomes consist of P1 (blue) and P2 (red) haploblocks. The maternally inherited embryo chromosomes consist of M1 (orange) and M2 (green) haploblocks. The haploblocks are assigned relative to the reference chromosomes.
Embryo in Detailed Haploblock Chart 	Individual informative SNPs are plotted in the detailed haploblock chart and colored according to the phase (M1, M2, P1, P2) that the SNP supports. • Key SNPs, which are strong evidence, are plotted above the haploblock bar. • Non-key SNPs, which are weaker evidence, are plotted below the haploblock bar. • Informative SNPs that are not successfully assigned a genotype (NC) are plotted in gray in the center of the haploblocks.

For users with the most common forms of color blindness, colors have been selected to aid discrimination between each pair of parental haplotypes. For further distinction between maternal and paternal haplotypes, P1 and P2 are colored without shading, whereas M1 and M2 are colored with shading.

References

1. Flicek P et al. (2013) Ensembl 2013. Nucleic Acids Res. 41 (Database issue). URL: http://www.ensembl.org/
2. Meyer LR, et al. (2013) The UCSC Genome Browser database: extensions and updates 2013. Nucleic Acids Res. 41. URL: http://genome.ucsc.edu
3. MacDonald JR, et al. The database of genomic variants: a curated collection of structural variation in the human genome. Nucleic Acids Res. 2013 Oct 29. URL: http://dgv.tcag.ca/dgv/app/home
4. Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD). URL: http://omim.org/
5. Gray, KA et al. (2011) Genenames.org: the HGNC resources in 2013. Nucleic Acids Res. 41 (Database issue).URL: http://www.genenames.org/
6. URL: http://decipher.sanger.ac.uk

   DECIPHER Data Display Agreement Notice

   These data are only available for display in the Browser, and not for bulk download. Access to bulk data may be obtained directly from DECIPHER (http://decipher.sanger.ac.uk/datasharing/) and is subject to a Data Access Agreement, in which the user certifies that no attempt to identify individual patients will be undertaken. The same restrictions apply to the public data displayed [at hosting institution] or [in this browser]: no one is authorized to attempt to identify patients by any means.

   This data is made available as soon as possible and may be a pre-publication release. For information on the proper use of DECIPHER data, please see http://decipher.sanger.ac.uk/datasharing. The DECIPHER consortium provides these data in good faith as a research tool, but without verifying the accuracy, clinical validity or utility of the data. The DECIPHER consortium, makes no warranty, express or implied, nor assumes any legal liability or responsibility for any purpose for which the data are used.
7. Data obtained from the ISCA Consortium database (www.iscaconsortium.org), which generates this information using study nstd37 in NCBI database of genomic structural variation (dbVAR, www.ncbi.nlm.nih.gov/dbvar). ISCA Consortium member laboratories provided samples and associated phenotype data.