

Sample (cell type, ID)	iPSC	BIHi257-A-2
Passage No.	47	
Bank ID	MB02	
DNA sample ID	B24_102	
Chip-ID and Position	208305090038, R11C01	
Date of testing	14.05.2024	
Call Rate	0.9919671	✓
Gender (provided/estimated from chip data)	Female	✓

Technology: Illumina BeadArray
Product: Illumina Infinium Global Screening Array-24 BeadChip
Manifest: GSAMD-24v3-0-EA_20034606_A1
Clusterfile: GSA-24v3-0_A1_ClusterFile

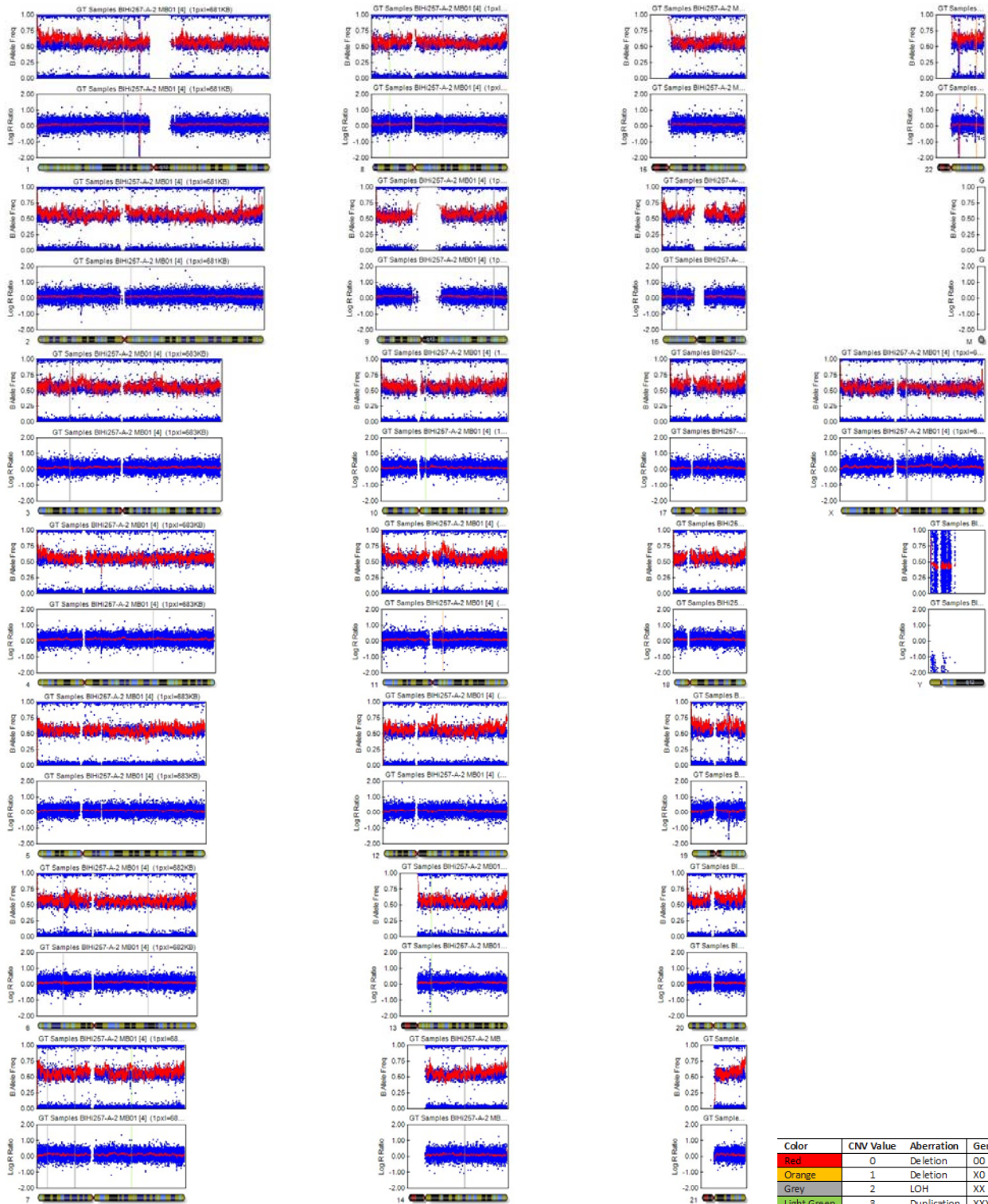
Genotype Analysis
GenomeStudio: GenomeStudio V2.0.5
Genotyping Module: V2.0.5

CNV Analysis
Algorithm: CNV-Partition
Version: 3.2.0

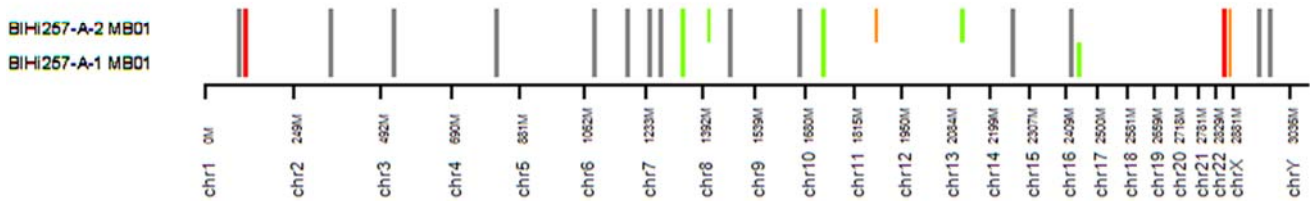
Parameters are set to detect copy number variations (CNVs) ≥ 45 kb and loss of heterozygosity (LOH) regions > 1 Mb with a confidence value > 35 . Balanced translocations and inversions cannot be detected with this method. Aberrant copy number regions are identified by log R ratio and B allele frequency. Copy number changes (gains and losses) greater than **0.4 Mb** and regions of LOH above **5 Mb** are considered reportable and taken into account for interpretation. Genomic positions are based on genome build GRCh37/hg19.

Virtual Karyotype

Gain (area marked in green), Loss (area marked in red), Loss of heterozygosity (area marked in gray)



Overview of detected CNV regions



Call Table

CNV regions found in BIHi257-A-2 MB01

Chr	Start	End	Size (bp)	CNV Value	Variant Type	Number of Genes*
1	92,155,337	93,521,909	1,366,572	2	LOH	
3	34,395,049	36,028,638	1,633,589	2	LOH	
6	118,335,317	119,460,786	1,125,469	2	LOH	
6	27,339,418	28,415,601	1,076,183	2	LOH	
7	39,796,229	41,277,798	1,481,569	2	LOH	
7	9,990,303	11,243,845	1,253,542	2	LOH	
7	100,886,307	101,142,529	256,222	3	Gain	
9	125,387,527	126,854,507	1,466,980	2	LOH	
10	47,074,201	47,703,869	629,668	3	Gain	10
X	70,468,754	72,779,929	2,311,175	2	LOH	

*Number of genes in CNV/LOH regions given only for **reportable** calls (see Appendix for details on genes in reported regions).

Interpretation

- There was **1** reportable copy number change identified in the BIHi257-A-2 MB01 MB01line.
 - > A 0,692 Mb Gain region on chromosome 10 was observed. 10 genes were found in this genomic region.

Further information about genes in detected regions and linked known diseases may be provided by the UCSC Genome Browser (<https://genome.ucsc.edu>) and Decipher (<https://decipher.sanger.ac.uk/search>).

Appendix

List of genes located in reportable CNV regions found in BIHi257-A-2 MB01

Gene	Chr	Transcription Start	Transcription End	Strand	Description
PPYR1	10	47,083,534	47,088,320	+	pancreatic polypeptide receptor 1
LOC72864 3	10	47,133,297	47,133,837	-	Homo sapiens cDNA clone IMAGE:5141022, partial cds.
ANXA8	10	47,157,986	47,174,123	-	annexin A8
ANXA8L1	10	47,157,991	47,174,041	-	annexin A8-like 1
FAM25G	10	47,177,224	47,181,689	-	
FAM25B	10	47,177,224	47,181,689	-	hypothetical protein LOC100132929
FAM25C	10	47,177,224	47,181,689	-	
LOC64282 6	10	47,242,687	47,243,495	-	Homo sapiens hypothetical protein LOC642826, mRNA (cDNA clone IMAGE:4296326).
FAM35A	10	47,379,720	47,421,237	+	Homo sapiens cDNA FLJ59018 complete cds, highly similar to Protein FAM35A.
ANTXRL	10	47,658,234	47,701,447	+	Homo sapiens cDNA FLJ32754 fis, clone TEST12001671.

References:

1. LaFramboise, T. (1 July 2009). "Single nucleotide polymorphism arrays: a decade of biological, computational and technological advances". *Nucleic Acids Research*. 37 (13): 4181–4193.
2. Arsham, M. S., Barch, M. J., & Lawce, H. J. (Eds.) (2017). *The AGT Cytogenetics Laboratory Manual (4th Ed.)*. Hoboken, NJ: John Wiley & Sons, Inc.
3. Haraksingh RR, Abyzov A, Urban AE. Comprehensive performance comparison of high-resolution array platforms for genome-wide Copy Number Variation (CNV) analysis in humans. *BMC Genomics*. 2017 Apr 24;18(1):321. doi: 10.1186/s12864-017-3658-x.
4. Wicell: <https://www.wicell.org/home/characterization/cytogenetics/snp-microarray/single-nucleotide-polymorphism-snp-mircroarray-.cmsx>