Single Nucleotide Polymorphism (SNP)- Karyotype

Sample (cell type, ID)	iPSC	BIHi257-A-1	
Passage No.	47		
Bank ID	MB02		
DNA sample ID	B24_101		
Chip-ID and Position	208305090038, R09C01		
Date of testing	14.05.2024		
Call Rate	0.9922225		**√**
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.

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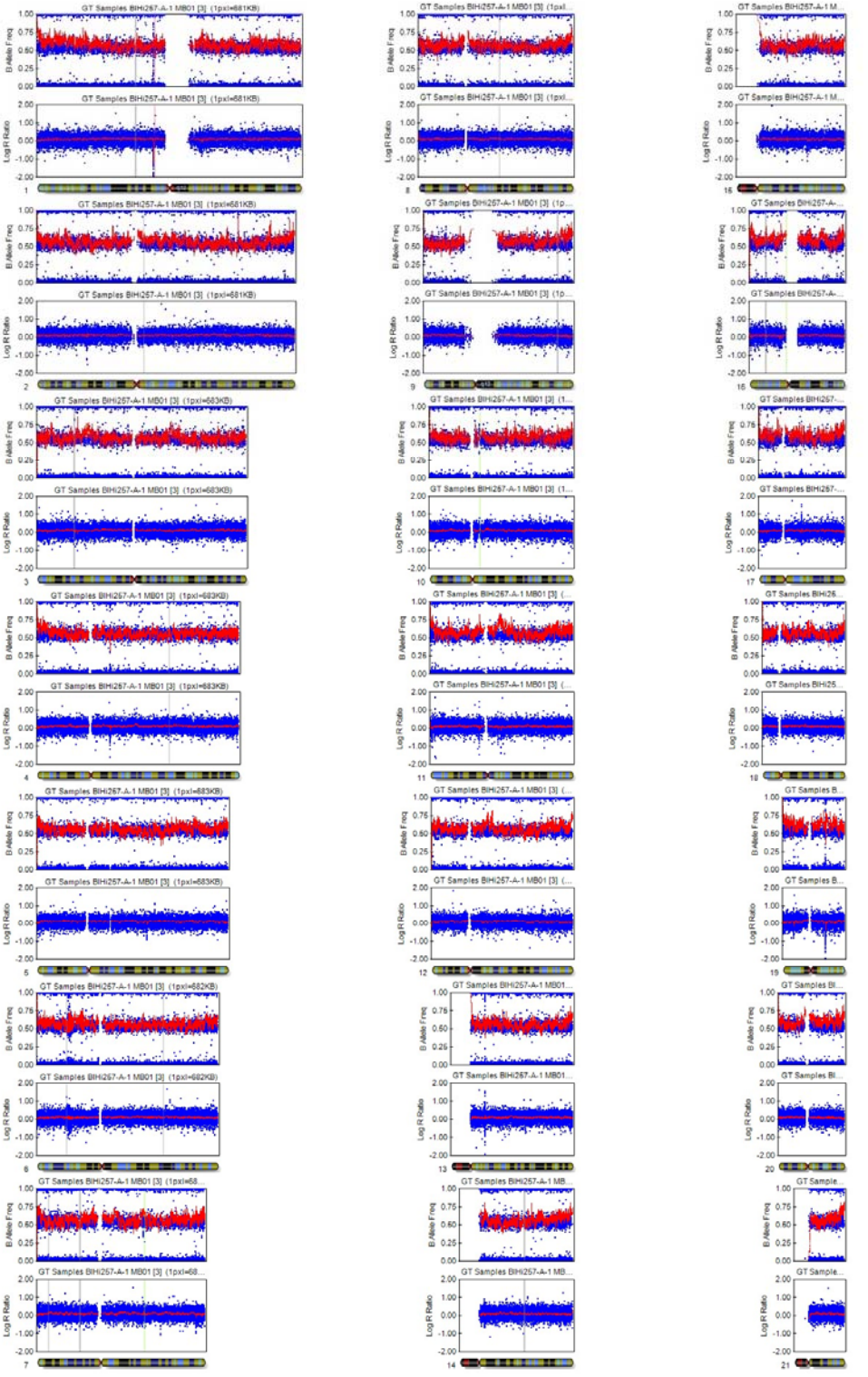
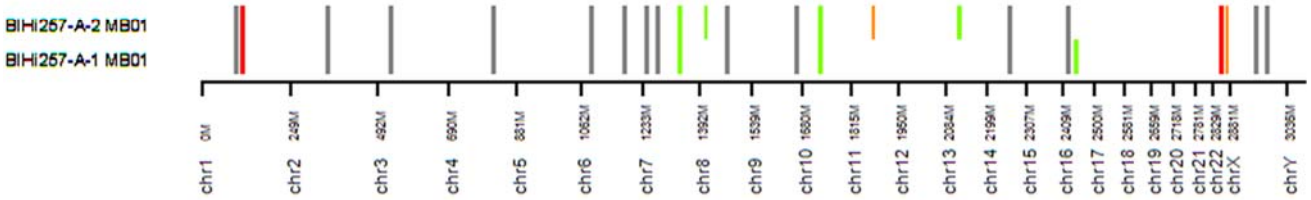
Virtual Karyotype

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



Color	CNV Value	Aberration	Genotype
Red	0	Deletion	00
Orange	1	Deletion	X0
Grey	2	LOH	XX
Light Green	3	Duplication	XXX
Dark Green	4	Duplication	XXXX

Overview of detected CNV regions



Call Table

CNV regions found in BIHi257-A-1 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	36,061,807	1,666,758	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	101,146,427	260,120	3	Gain	
9	125,387,527	126,854,507	1,466,980	2	LOH	
10	**47,011,498**	**47,703,869**	**692,371**	**3**	**Gain**	**10**
16	34,428,972	34,732,946	303,974	3	Gain	
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-1 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-1 MB01

Gene	Chr	Transcription Start	Transcription End	Strand	Description
PPYR1	10	47,083,534	47,088,320	+	pancreatic polypeptide receptor 1
LOC728643	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	-	
LOC642826	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 TESTI2001671.

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