

Karyotype Report

Customer sample ID: hvs540c
 Internal sample ID: NL96GSAUMCD100008

Gender
 Stated by customer: Female
 According to array data: Female

Copy Number Analysis

Copy number events are reported when they exceed 50 kb in size, have a confidence value greater than 50, and are confirmed by visual inspection of the B-allele frequency (BAF) and log R ratio profiles. CNVs located in known hot spot regions of recurrent mutations in hPSCs are additionally highlighted in the summary table. Loss of heterozygosity (LOH) are reported for regions larger than 1 Mbp.

The reported CNVs can be checked against the [Database of Genomic Variants](#) which provides a comprehensive summary of structural variation in the human genome. The content of the database only includes structural variations identified in healthy control samples.

Affected genes can be monitored by using a genome browser such as the NCBI [Genome Data Viewer](#), [Ensembl Genome Browser](#) or the [UCSC Genome Browser](#).

SampleID	Chr	Start	End	Size	Value	Hotspot region	LOH region
NL96GSAUMCD100008 [5]	2	142152653	142228509	75856	0	no	no
NL96GSAUMCD100008 [5]	4	69405882	69512937	107055	0	no	no
NL96GSAUMCD100008 [5]	8	4522427	4750206	227779	1	no	no
NL96GSAUMCD100008 [5]	11	103014125	103153788	139663	1	no	no
NL96GSAUMCD100008 [5]	12	88454677	88535064	80387	1	no	no
NL96GSAUMCD100008 [5]	13	48877851	49050882	173031	1	no	no
NL96GSAUMCD100008 [5]	16	28634373	29044745	410372	1	yes	no
NL96GSAUMCD100008 [5]	17	33681007	33766040	85033	1	no	no
NL96GSAUMCD100008 [5]	17	71833923	72691941	858018	3	no	no
NL96GSAUMCD100008 [5]	19	49027683	49116359	88676	3	no	no

Conclusion: The sample shows duplications on chr 17 and 19 as well as copy losses on chr 2 (only one allele, Value: 0 is wrong interpretation of software), 4 (here: both alleles), 8, 11, 12, 13, 16 and 17 with the lengths specified in the table.

Karyogram





Karyotyping

Technical annex

Technology used: Illumina BeadArray

Product: Global Screening Array + Multi Disease content 24
v3 BeadChip

Manifest file: GSAMD-24v3-0-EA_20034606_A1.bpm
Cluster file: GSAMD24v3-0-EA_20034606_A1.egt

Chip barcode and segment: 209838860068 R09C01

Batch ID and 96 well position: WG7941481-MSA3 E01

Call rate: 0.9900679

Typing

Scanner: Illumina iScan, S/N: N263
Site of processing: Life&Brain GENOMICS, Bonn, Germany
Manufacturer: Illumina, Inc., San Diego, United States of America

Genotype Analysis

Genome Studio: GenomeStudio V2.0.5
Genotyping module: Vers. 2.0.5

Copy Number Analysis

Algorithm applied: cnvPartition
Version: 3.2
Software producer: Illumina, Inc., San Diego, United States of America

Analyst

Dr. Michael Peitz
mpeitz@lifeandbrain.com
Tel.: +49 228 6885 156