# Karyotype Report

Customer sample ID: hvs540a
Internal sample ID: NL53GSAUMCD100006

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.

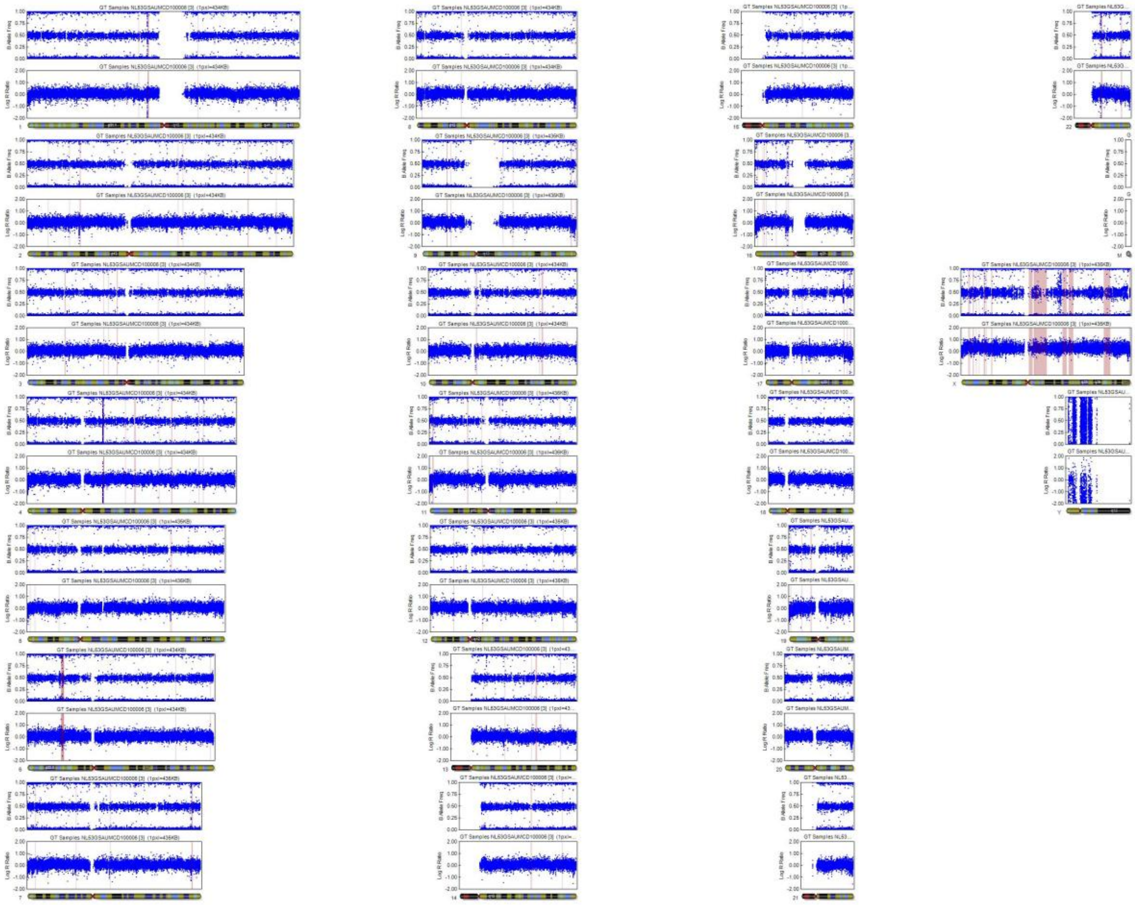
The reported CNVs can be checked against the Database of Genomic Variants which provides a comprehensive summary of structural variation in the human genome. The content of the database only includes structural variations identified in healthy control samples.

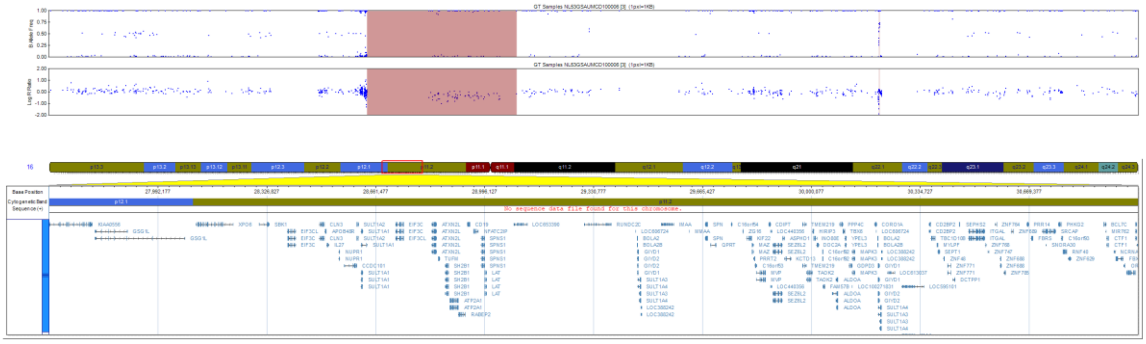
Affected genes can be monitored by using a genome browser such as the NCBI Genome Data Viewer, Ensembl Genome Browser or the UCSC Genome Browser.

| SampleID | Chr | Start | End | Size | Value | Hotspot region | LOH region |
|---|---|---|---|---|---|---|---|
| NL53GSAUMCD100006 [3] | 2 | 142152653 | 142228509 | 75856 | 1 | no | no |
| NL53GSAUMCD100006 [3] | 4 | 69418832 | 69512937 | 94105 | 0 | no | no |
| NL53GSAUMCD100006 [3] | 8 | 4522427 | 4750206 | 227779 | 1 | no | no |
| NL53GSAUMCD100006 [3] | 16 | 28634373 | 29095150 | 460777 | 1 | yes | no |
| NL53GSAUMCD100006 [3] | 16 | 88491482 | 88601360 | 109878 | 1 | yes | no |
| NL53GSAUMCD100006 [3] | 17 | 33681007 | 33766040 | 85033 | 1 | no | no |
| NL53GSAUMCD100006 [3] | 17 | 71833923 | 72688818 | 854895 | 3 | no | no |

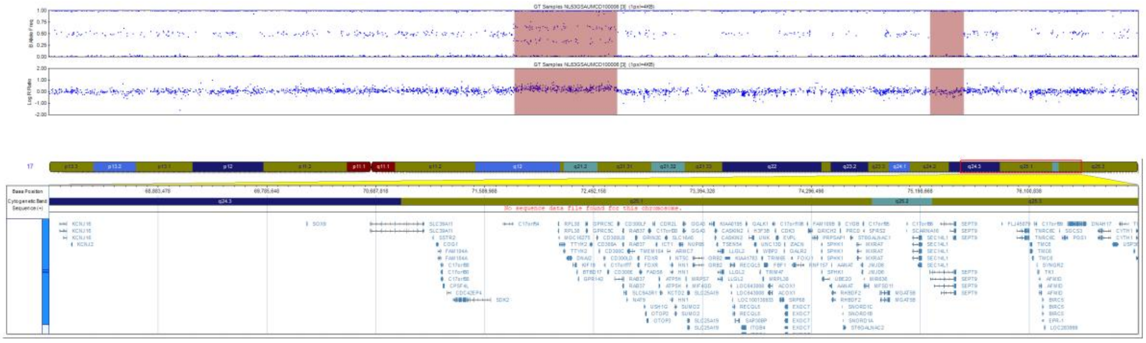
**Conclusion**: The sample shows a duplication on chr 17 and copy losses on chr 2, 4 (here: both alleles), 8, 16 and 17 with the lengths specified in the table.

# Karyogram





NL53GSAUMCD100006
hvs540a



## 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 R05C01

Batch ID and 96 well position: WG7941481-MSA3 C01

Call rate: 0.9906569

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

NL53GSAUMCD100006
hvs540a