

## Karyotype Report

Customer sample ID: hvs520a  
 Internal sample ID: NL80GSAUMCD100005

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.

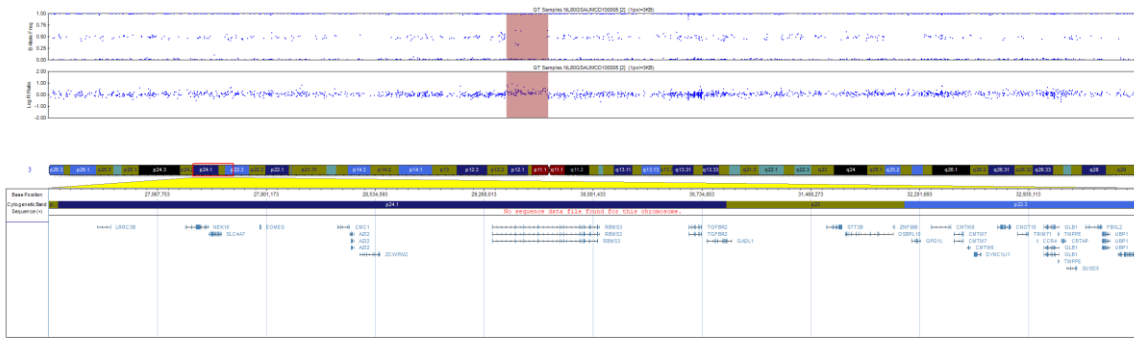
Affected genes can be monitored by using a genome browser such as the NCBI [Genome Data Viewer](#), [Ensembl Genome Browser](#) or the [UCSC Genome Browser](#).

SampleID	Chr	Start	End	Size	Value	Hotspot region	LOH region
NL80GSAUMCD100005 [2]	1	230521737	232423446	1901709	2	no	yes
NL80GSAUMCD100005 [2]	3	81414970	84770561	3355591	2	no	yes
NL80GSAUMCD100005 [2]	3	29418596	29700225	281629	3	no	no
NL80GSAUMCD100005 [2]	7	126429517	127915376	1485859	2	no	yes

**Conclusion:** The sample shows a duplication on chr 3 and LOH regions on chr 1, 3 and 7 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 R03C01

Batch ID and 96 well position: WG7941481-MSA3 B01

Call rate: 0.9910706

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

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