

## Karyotype Report

Customer sample ID: hvs556a  
Internal sample ID: NL40GSAUMCD100046

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

## 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
NL40GSAUMCD100046 [43]	1	1203938	1289863	85925	1	no	no
NL40GSAUMCD100046 [43]	4	30296776	31399076	1102300	2	no	yes
NL40GSAUMCD100046 [43]	7	4794220	4857977	63757	1	no	no
NL40GSAUMCD100046 [43]	12	111883779	113944788	2061009	2	no	yes
NL40GSAUMCD100046 [43]	16	1782964	1860360	77396	1	yes	no

**Conclusion:** The sample shows copy losses on chr 1, 7 and 16 as well as LOH regions on chr 4 and 12 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: 209838860171 R02C02

Batch ID and 96 well position: WG7941481-MSA3 C06

Call rate: 0.99541

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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