

## Karyotype Report

Customer sample ID: hvs556b  
Internal sample ID: NL13GSAUMCD100047

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
NL13GSAUMCD100047 [44]	4	30296776	31399076	1102300	2	no	yes
NL13GSAUMCD100047 [44]	12	111296418	113944788	2648370	2	no	yes

**Conclusion:** The sample shows LOH regions on chr 4 and 12 with the lengths specified in the table.

# Karyogram



NL13GSAUMCD100047  
hvs556b

## 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 R04C02

Batch ID and 96 well position: WG7941481-MSA3 D06

Call rate: 0.9958017

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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