Karyotype Report

Customer sample ID: hvs556c
Internal sample ID: NL83GSAUMCD100048

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
NL83GSAUMCD100048 [45]	4	30296776	31399076	1102300	2	no	yes
NL83GSAUMCD100048 [45]	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

NL83GSAUMCD100048
hvs556c

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 R06C02
Batch ID and 96 well position:	WG7941481-MSA3 E06
Call rate:	0.9956921

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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