Karyotype Report

Customer sample ID: hvs576b
Internal sample ID: NL74GSAUMCD100016

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.

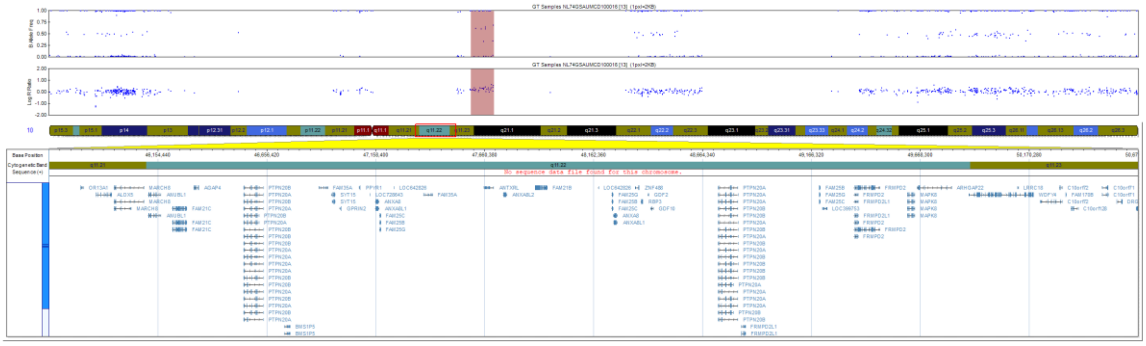
The reported CNVs can be checked against the Database of Genomic Variants which provides a comprehensive summary of structural variation in the human genome. The content of the database only includes structural variations identified in healthy control samples.

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
NL74GSAUMCD100016 [13]	8	96797851	99092060	2294209	2	yes	yes
NL74GSAUMCD100016 [13]	10	47596804	47703869	107065	3	no	no
NL74GSAUMCD100016 [13]	12	88454677	88566450	111773	1	no	no
NL74GSAUMCD100016 [13]	13	48877851	49047531	169680	1	no	no

Conclusion: The sample shows a duplication on chr 10 and copy losses on chr 12 and 13 as well as an LOH region on chr 8 with the lengths specified in the table.

Karyogram



NL74GSAUMCD100016
hvs576b

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 R01C02

Batch ID and 96 well position: WG7941481-MSA3 E02

Call rate: 0.9907624

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