

Karyotype Report

Customer sample ID: hvs577a
Internal sample ID: NL20GSAUMCD100018

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.

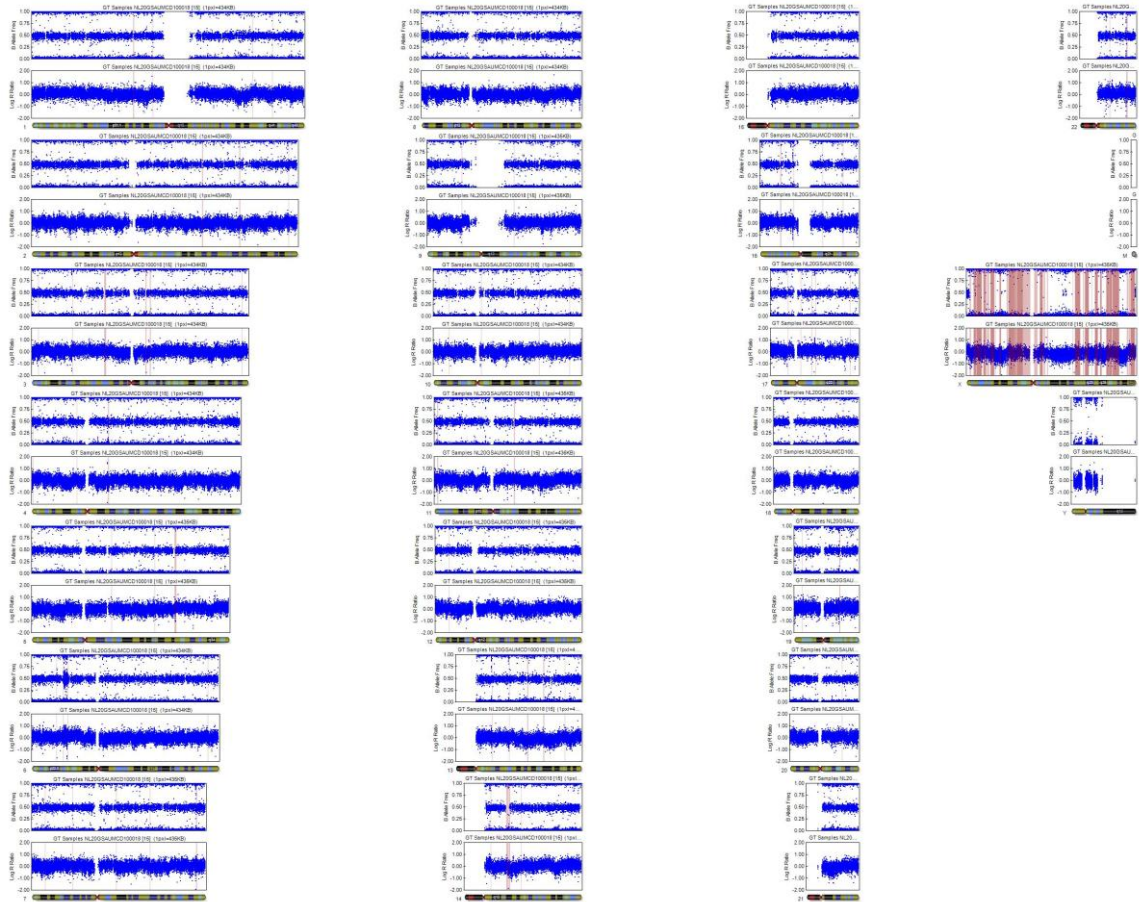
The reported CNVs can be checked against the [Database of Genomic Variants](#) which provides a comprehensive summary of structural variation in the human genome. The content of the database only includes structural variations identified in healthy control samples.

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
NL20GSAUMCD100018 [15]	3	65797540	67925920	2128380	2	no	yes
NL20GSAUMCD100018 [15]	14	38443309	40543226	2099917	2	yes	yes

Conclusion: The sample shows LOH regions on chr 3 and 14 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 R05C02

Batch ID and 96 well position: WG7941481-MSA3 G02

Call rate: 0.9954428

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