Karyotype Report

Customer sample ID: hvs578a
Internal sample ID: NL36GSAUMCD100021

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.

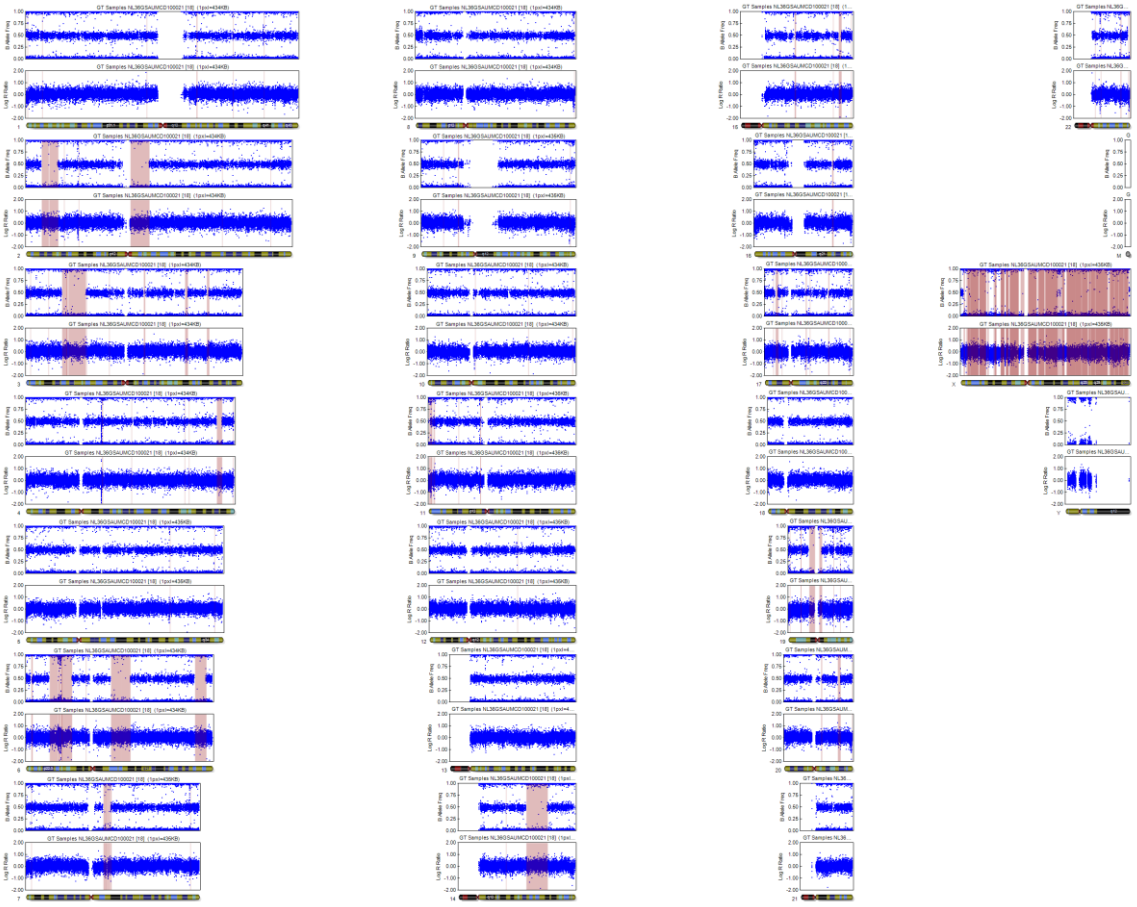
Affected genes can be monitored by using a genome browser such as the NCBI Genome Data Viewer, Ensembl Genome Browser or the UCSC Genome Browser.

SampleID	Chr	Start	End	Size	Value	Hotspot region	LOH region
NL36GSAUMCD100021 [18]	1	1203938	1289863	85925	1	no	no
NL36GSAUMCD100021 [18]	1	189211031	189353426	142395	1	no	no
NL36GSAUMCD100021 [18]	1	217487940	217770874	282934	3	no	no
NL36GSAUMCD100021 [18]	2	14303988	21225269	6921281	2	no	yes
NL36GSAUMCD100021 [18]	2	21266783	29667972	8401189	2	no	yes
NL36GSAUMCD100021 [18]	2	95345619	113082042	17736423	2	no	yes
NL36GSAUMCD100021 [18]	3	33035529	37038193	4002664	2	no	yes
NL36GSAUMCD100021 [18]	3	38645439	55109175	16463736	2	no	yes
NL36GSAUMCD100021 [18]	3	145492901	148345526	2852625	2	no	yes
NL36GSAUMCD100021 [18]	4	174776773	179813733	5036960	2	no	yes
NL36GSAUMCD100021 [18]	4	69401172	69512637	111465	0	no	no
NL36GSAUMCD100021 [18]	4	70157231	70219930	62699	0	no	no
NL36GSAUMCD100021 [18]	4	144925824	145033810	107986	3	no	no
NL36GSAUMCD100021 [18]	6	4605824	6171892	1566068	2	no	yes
NL36GSAUMCD100021 [18]	6	21684733	29855978	8171245	2	no	yes
NL36GSAUMCD100021 [18]	6	30855907	32452443	1596536	2	no	yes
NL36GSAUMCD100021 [18]	6	32596393	42407020	9810627	2	no	yes
NL36GSAUMCD100021 [18]	6	77624386	87251194	9626808	2	no	yes
NL36GSAUMCD100021 [18]	6	87267246	95880490	8613244	2	no	yes
NL36GSAUMCD100021 [18]	6	154463406	164925833	10462427	2	no	yes
NL36GSAUMCD100021 [18]	7	70995459	75613147	4617688	2	no	yes
NL36GSAUMCD100021 [18]	7	76871048	78258818	1387770	2	no	yes
NL36GSAUMCD100021 [18]	11	308180	2238886	1930706	2	no	yes
NL36GSAUMCD100021 [18]	12	80546967	80655816	108849	3	no	no
NL36GSAUMCD100021 [18]	14	62037479	81919396	19881917	2	yes	yes
NL36GSAUMCD100021 [18]	15	89448201	92315474	2867273	2	no	yes
NL36GSAUMCD100021 [18]	17	9872446	12531983	2659537	2	no	yes

NL36GSAUMCD100021 [18]	19	18922444	24572955	5650511	2	no	yes
NL36GSAUMCD100021 [18]	19	28295716	30973192	2677476	2	no	yes
NL36GSAUMCD100021 [18]	20	49197873	52165215	2967342	2	no	yes

Conclusion: The sample shows copy losses on chr 1 and 4 (here: both copies) as well as duplications on chr 1 and 4 along with several LOH regions ("Value: 2") on various chromosomes with the lengths specified in the table.

Karyogram



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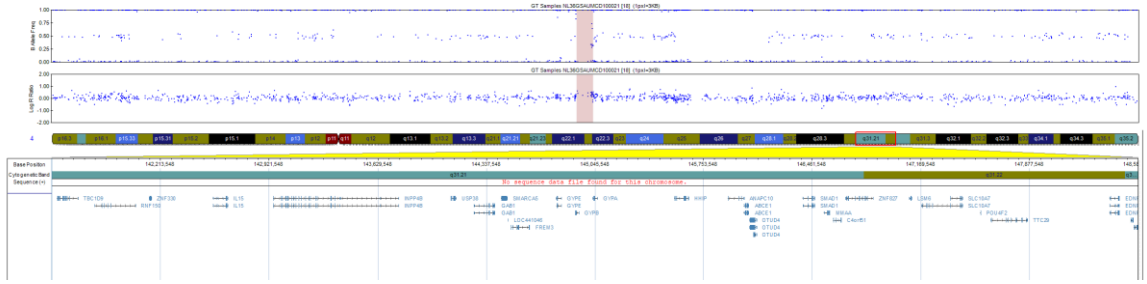

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Karyotyping
Technical annex

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 R11C02
Batch ID and 96 well position:	WG7941481-MSA3 B03
Call rate:	0.9954401

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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