Copy number variation (CNV) analysis

Document No.	2015-06
Issue date	2018-10-15

Stem cell Information

Stem cell line	**hUSiPS2**	Institute	KSCR
Cell type	**hiPSC**	Inspected Date	
Banking status	**MCB**	Issue date	2018-10-15
Passage	**p25**		
Note			

Experiment type

SNP chip

Platform	illumina Human Omni 2.5 Exome Beadchip	**Analysis program**	GenomeStudio, PennCNV
Reference	hg19	**Analysis document**	SOP#26-Ver.4

Statistics

	Total	Gain	Loss	
the number of total CNVs	13	8	5	
the number of manually filtered CNVs	5	4	1	
the number of CNVs excluded Korean normal CNV DB (KGVDB)	4	3	1	

Result of Data Analysis

List of CNVs

	Total	Gain	Loss	Cytoband
The number of total CNV calls	**5**	**4**	**1**	
The number of Pathogenic CNVs	3	3	0	7q36.2, 16p13.13, 20q11.21
The number of Recurrent CNVs	1	1	0	20q11.21
The number of stemness-related CNVs	1	1	0	20q11.21
The number of Differentiation-related CNVs	1	1	0	20q11.21
The number of cancer-related CNVs	2	2	0	16p13.13, 20q11.21
The number of immunogenicity-related CNVs	1	1	0	20q11.21

*Recurrent CNVs include CNV gain on 1q41, 12p13.31, 17q25.2 and 20q11.21, CNV loss on 10p11.22

Interpretation

Copy number variation (CNV) analysis

Document	2015-06
Issue date	2018-10-15

Stem cell Information

Stem cell line	hUSiPS2	Institute	KSCR
Cell type	hiPSC	Inspected	
Banking status	MCB	Issue date	2018-10-15
Passage	p25		
Note			

Result of Data Analysis

List of CNVs

chr	Cytoband	chr_start	chr_end	length	Copy Number	CNV	Genes	Pathogenic CNV	Phenotype	Recurrent CNV	Cancer-related	Stemness-related	Differentiation-related	immunogenicity-related	KGVDB
7	q36.1	149,551,993	149,666,820	114,828	cn=3	Gain	ATP6V0E2,ATP6V0E2-AS1,ZNF862	.	.	.	.	.	.	.	No
7	q36.2	153,522,051	153,660,141	138,091	cn=3	Gain	DPP6	DPP6	Mental retardation (616311), Ventricular	.	.	.	.	.	Yes
15	q11.2	22,410,067	22,512,696	102,630	cn=1	Loss	OR4N3P	.	.	.	.	.	.	.	No
16	p13.13	10,754,456	11,024,105	269,650	cn=3	Gain	CIITA,DEXI,NUBP1,TEKT5,TVP23A	CIITA	Bare lymphocyte syndrome typeII (209920), Rheumatoid	.	CIITA	.	.	.	No
20	q11.21	29,804,016	31,640,095	1,836,080	cn=3	Gain	ABALON,ASXL1,BCL2L1,BPIFB2,BPIFB6,C20orf203,CCM2L,COMMD7,COX4I2,DEFB115,DEFB116,DEFB118,DEFB119,DEFB121,DEFB122,DEFB123,DEFB124,DNMT3B,DUSP15,FOXS1,HCK,HM13,HM13-AS1,ID1,KIF3B,LINC00028,LOC101929698,LOC149950,MAPRE1,MCTS2P,MIR1825,MIR3193,MIR7641-2,MYLK2,NOL4L,PDRG1,PLAGL2,POFUT1,REM1,SUN5,TM9SF4,TPX2,TSPY26P,TTLL9,XKR7	COX4I2, DNMT3B, MYLK2, ASXL1	COX4I2 (Exocrine pancreatic insufficiency) DNMT3B(Immonudeficiency-centromeric instability-facial anomalies syn I) MYLK2 (Cardiomyopathy) ASXL1 (Bohring-Opitz syn, Myelodysplastic syn) SNTA1(LongQT syn)	Yes	ASXL1, BCL2L1	DNMT3B	DMMT3B	DEFB119, DEFB123	No

질병관리본부