Copy number variation (CNV) analysis

Document No.	2019-12
Issue date	2019-12-05

Stem cell Information

Stem cell line	**hFSiPS3-1**	Institute	KSCR
Cell type	**hiPSC**	Inspection date	2019-09-30
Banking status	**Cell line distribution for research**	Issue date	2019-12-05
Passage	**p30**		
Note			

Experiment type

SNP chip

Platform	Affymetrix CytoscanHD	**Analysis program**	ChAS 4.0
Reference	hg38	**Analysis document**	SOP#26.1-Ver.5

Statistics

	Total	Gain	Loss	
the number of total CNVs	2			
the number of manually filtered CNVs	1	1	0	

Result of Data Analysis

List of CNVs

	Total	Gain	Loss	Cytoband
The number of total CNV calls	**1**	**1**	**0**	**20q11.21**
The number of Pathogenic CNVs	1	1	0	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	1	1	0	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

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Result of Data Analysis

List of CNVs

chr	Cytoband	chr_start	chr_end	length (kbp)	Copy Number	CNV	Genes	OMIM Gene counts	OMIM genes (Phenotype)	Recurrent CNV	Cancer-related	Stemness-related	Differentiation-related	immunog enicity-related
20	q11.21	31,245,731	32,969,277	1723.5	3	Gain	DEFB115, DEFB116, DEFB118, DEFB119, DEFB121, DEFB122, DEFB123, DEFB124, REM1, LINC00028, HM13, HM13- AS1, ID1, MIR3193, COX4I2, BCL2L1, ABALON, TPX2, MYLK2, FOXS1, DUSP15, TTLL9, PDRG1, XKR7, MIR7641-2, CCM2L, HCK, TM9SF4, TSPY26P, PLAGL2, POFUT1, MIR1825, KIF3B, ASXL1, NOL4L, LOC101929698, LOC149950, C20orf203, COMMD7, DNMT3B,	24	DEFB118 (607650), DEFB119 (615997), DEFB121 (616075), DEFB122 (616077), DEFB123 (616076), REM1 (610388), HM13 (607106), ID1 (600349), COX4I2 (607976), BCL2L1 (600039), ABALON (616018), TPX2 (605917), MYLK2 (606566), FOXS1 (602939), DUSP15 (616776), PDRG1 (610789), HCK (142370), PLAGL2 (604866), POFUT1 (607491), KIF3B (603754), ASXL1 (612990), COMMD7 (616703), DNMT3B (602900), MAPRE1 (603108)	Yes	BCL2L1, AS	DNMT3	DNMT3	DEFB119, DEFB123