Copy number variation (CNV) analysis

Document No.	2015-04
Issue date	2018-10-15

Stem cell Information

Stem cell line	hFmiPS2	Institute	KSCR
Cell type	hiPSC	Inspected Date	
Banking status	DCB	Issue date	2018-10-15
Passage	p27		
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	8	6	2	
the number of manually filtered CNVs	6	5	1	
the number of CNVs excluded Korean normal CNV DB (KGVDB)	6	5	1	

Result of Data Analysis

List of CNVs

	Total	Gain	Loss	Cytoband
The number of total CNV calls	**6**	**5**	**1**	
The number of Pathogenic CNVs	3	3	0	4q31.21, 20q11.21, 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	2	2	0	4q31.21, 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	Copy Number	CNV
1	q21.3	151,376,067	151,408,434	32,368	cn=3	Gain
4	**q31.21**	**144,990,400**	**145,035,788**	**45,389**	**cn=3**	**Gain**
11	q25	131,511,581	131,605,278	93,698	cn=1	Loss
20	q11.21	29,863,685	29,909,618	45,934	cn=3	Gain
20	**q11.21**	**30,021,789**	**30,347,832**	**326,044**	**cn=3**	**Gain**
20	**q11.21**	**30,681,285**	**32,806,891**	**2,125,607**	**cn=3**	**Gain**