

Copy number variation (CNV) analysis			Document No.	2014-06
			Issue date	2018-10-15
Stem cell Information				
Stem cell line	hFmiPS1	Institute	KSCR	
Cell type	hiPSC	Inspection date		
Banking status	DCB	Issue date	2018-10-15	
Passage	p30			
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	14	9	5	
the number of manually filtered CNVs	7	4	3	
the number of CNVs excluded Korean normal CNV DB (KGVDB)	7	4	3	
Result of Data Analysis				
List of CNVs				
	Total	Gain	Loss	Cytoband
The number of total CNV calls	7	4	3	
The number of Pathogenic CNVs	2	2	0	1q21.3, 4q31.21
The number of Recurrent CNVs	.	.	.	
The number of stemness-related CNVs	.	.	.	
The number of Differentiation-related CNVs	.	.	.	
The number of cancer-related CNVs	1	1	0	1q21.3
The number of immunogenicity-related CNVs	1	1	0	4q31.21
*Recurrent CNVs include CNV gain on 1q41, 12p13.31, 17q25.2 and 20q11.21, CNV loss on 10p11.22				
Interpretation				

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Passage	p30		
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 - relate	Stemness-related	Differentiation-related	immuno genicity-related	KGVDB
1	p21.1	106,553,138	108,542,462	1,989,325	cn = 1	Loss	MIR7852,NTN G1,PRMT6,VAV 3,VAV3-AS1	No
1	q21.3	150,490,849	150,571,730	80,882	cn = 3	Gain	ADAMTSL4, ADAMTSL4-AS1,MCL1,MI R4257	ADAMTSL4	Ectopia lentis et pupillae (225200), Ectopia	.	MCL1	.	.	.	No
1	q21.3	151,375,584	151,408,434	32,851	cn = 3	Gain	POGZ	No
4	q31.21	145,020,490	145,039,259	18,770	cn = 3	Gain	GYPA	GYPA	Malaria (611162)	GYPA	No
4	q35.2	188,945,045	189,086,300	141,256	cn = 3	Gain	TRIML1,TRIML2	No
10	p11.1	38,774,749	39,154,535	379,787	cn = 1	Loss	ACTR3BP5	No
11	q25	131,511,581	131,605,278	93,698	cn = 1	Loss	NTM-AS1	No