Copy number variation (CNV) analysis

Document No.	2014-02
Issue date	2018-10-15

Stem cell Information

Stem cell line	SNUhES31	Institute	KSCR
Cell type	hESC	Inspection date	
Banking status	DCB	Issue date	2018-10-15
Passage	p34		
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	56	7	49	
the number of manually filtered CNVs	31	3	28	
the number of CNVs excluded Korean normal CNV DB (KGVDB)	29	3	26	

Result of Data Analysis

List of CNVs

	Total	Gain	Loss	Cytoband
The number of total CNV calls	**31**	**3**	**28**	
The number of Pathogenic CNVs	11	0	11	1p36.33, 4p16.3, 7p22.1, 8q24.3, 8q24.3, 9q34.3, 11p15.5, 11p15.5, 16p13.3, 21q22.3, 22q13.33
The number of Recurrent CNVs	.	.	.	
The number of stemness-related CNVs	.	.	.	
The number of Differentiation-related CNVs	.	.	.	
The number of cancer-related CNVs	2	0	2	9q34.3, 16p13.3
The number of immunogenicity-related CNVs	3	0	3	1p36.33, 11p15.5, 21q22.3

**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	Genes	Pathogenic CNV	Phenotype	Recurrent CNV	Cancer-related	Stemness-related	Differentiatio	immunogenicity-related	KGVDB
1	**p36.33**	**894,573**	**1,146,696**	**252,124**	**cn=1**	**Loss**	**AGRN,C1orf159,HES4,ISG15,KLHL17,LINC01342,MIR2**	**AGRN**	**Myasthenic syndrome (615120)**	.	.	.	.	TNFRSF18	No
1	p36.21	13,176,405	13,189,898	13,494	cn=3	Gain	HNRNPCL2	.	.	.	.	.	.	.	No
2	q37.3	241,567,923	241,577,705	9,783	cn=1	Loss	GPR35	.	.	.	.	.	.	.	No
4	**p16.3**	**2,798,170**	**2,826,400**	**28,231**	**cn=1**	**Loss**	**SH3BP2**	**SH3BP2**	**Cherubism (118400)**	.	.	.	.	.	No
5	p13.3	32,106,628	32,156,633	50,006	cn=3	Gain	GOLPH3,PDZD2	.	.	.	.	.	.	.	No
7	p22.3	943,381	1,064,525	121,145	cn=1	Loss	ADAP1,C7orf50,COX19,CYP2	.	.	.	.	.	.	.	No
7	p22.3	1,504,407	1,526,300	21,894	cn=1	Loss	INTS1	.	.	.	.	.	.	.	No
7	**p22.1**	**4,822,887**	**4,843,868**	**20,982**	**cn=1**	**Loss**	**AP5Z1,MIR4656,RADIL**	**AP5Z1**	**Spastic paraplegia 48 (613647)**	.	.	.	.	.	No
8	**q24.3**	**144,803,220**	**144,809,006**	**5,787**	**cn=1**	**Loss**	**FAM83H,MAPK15**	**FAM83H**	**Amelogenesis imperfecta type III (130900)**	.	.	.	.	.	Yes
8	**q24.3**	**144,993,975**	**145,025,515**	**31,541**	**cn=1**	**Loss**	**MIR661,PLEC**	**PLEC**	**Epidermolysis bullosa (616487, 226670, 612138, 131950), Muscular dystrophy, type 2Q**	.	.	.	.	.	No
9	q34.2	137,265,069	137,278,394	13,326	cn=1	Loss	MIR4669,RXRA	.	.	.	.	.	.	.	No
9	q34.3	139,218,885	139,254,317	35,433	cn=1	Loss	DKFZP434A062,GPSM1	.	.	.	.	.	.	.	No
9	**q34.3**	**139,405,093**	**139,412,197**	**7,105**	**cn=1**	**Loss**	**NOTCH1**	**NOTCH1**	**Adams-Oliver syndrome 5 (616028), Aortic valvu disease 1 (109730)**	.	NOTCH1	.	.	.	No
11	**p15.5**	**1,250,488**	**1,431,083**	**180,596**	**cn=1**	**Loss**	**BRSK2,MIR6744,MUC5B,T**	**MUC5B**	**Pulmonary fibrosis (178500)**	.	.	.	.	TOLLIP	Yes
11	**p15.5**	**2,179,313**	**2,192,798**	**13,486**	**cn=1**	**Loss**	**INS,INS-IGF2,TH**	**INS, TH**	**INS [Diabetes mellitus (125852, 606176), Hyperproinsulinemi a (616214), Maturity-onset disease of the young (613370)] ; TH**	.	.	.	.	.	No
11	q13.1	64,661,532	64,685,482	23,951	cn=1	Loss	ATG2A,MIR6749,MIR6750	.	.	.	.	.	.	.	No
12	q24.31	123,465,877	123,484,647	18,771	cn=1	Loss	ARL6IP4,PITPNM2	.	.	.	.	.	.	.	No
14	q32.33	105,609,836	105,644,421	34,586	cn=1	Loss	JAG2,MIR6765,NUDT14	.	.	.	.	.	.	.	No
14	q32.33	105,941,933	105,965,215	23,283	cn=1	Loss	C14orf80,CRIP1,CRIP2	.	.	.	.	.	.	.	No
16	p13.3	661,142	696,111	34,970	cn=1	Loss	C16orf13,FAM195A,RAB40C,	.	.	.	.	.	.	.	No
16	p13.3	733,947	746,914	12,968	cn=1	Loss	FBXL16,JMJD8,WDR24	.	.	.	.	.	.	.	No
16	p13.3	1,139,116	1,176,214	37,099	cn=1	Loss	C1QTNF8	.	.	.	.	.	.	.	No
16	**p13.3**	**2,132,571**	**2,159,242**	**26,672**	**cn=1**	**Loss**	**MIR1225,MIR6511B1,MIR6511B2,PKD1,TSC2**	**TSC2, PKD1**	**TSC2 [Lymphangioleiomyomatosis (606690), Tuberous sclerosis-2 (613254)] ; PKD1[Polycystic kidney disease**	.	TSC2	.	.	.	No
17	q25.3	79,506,356	79,518,598	12,243	cn=1	Loss	FAAP100	.	.	.	.	.	.	.	No
19	p13.3	1,003,109	1,020,409	17,301	cn=1	Loss	GRIN3B,TMEM259	.	.	.	.	.	.	.	No
19	p13.3	1,806,567	1,826,112	19,546	cn=1	Loss	ATP8B3,LOC100288123,MIR	.	.	.	.	.	.	.	No
19	p13.3	3,611,765	3,624,838	13,074	cn=1	Loss	CACTIN,CACTIN-AS1	.	.	.	.	.	.	.	No
19	p13.3	4,028,067	4,044,579	16,513	cn=1	Loss	PIAS4,ZBTB7A	.	.	.	.	.	.	.	No
20	q11.21	29,584,541	29,629,342	44,802	cn=3	Gain	FRG1BP,FRG1DP	.	.	.	.	.	.	.	No
21	**q22.3**	**46,319,417**	**46,325,126**	**5,710**	**cn=1**	**Loss**	**ITGB2**	**ITGB2**	**Leukocyte adhesion dificiency (116920)**	.	.	.	.	ITGB2	No

22	q13.33	50,957,520	50,963,464	5,945	cn=1	Loss	NCAPH2,SCO2	SCO2	Cardioencephalomyopathy (604377). Myopia 6 (608908)	.	.	.	.	.	No