

Copy number variation (CNV) analysis

Document No.	2016-04
Issue date	2018-10-15

Stem cell Information

Stem cell line	CHAhES15	Institute	KSCR
Cell type	hESC	Inspection date	
Banking status	PCB	Issue date	2018-10-15
Passage	p28		
Note	cultured on matrigel		

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	154	153	1	
the number of manually filtered CNVs	10	9	1	
the number of CNVs excluded Korean normal CNV DB (KGVDB)	9	8	1	

Result of Data Analysis

List of CNVs

	Total	Gain	Loss	Cytoband
The number of total CNV calls	10	9	1	
The number of Pathogenic CNVs	2	2	0	1q21.2, 7q36.2
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	1q21.2
The number of immunogenicity-related CNVs	1	1	0	1q21.2

*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	Differentiation-related	immunogenicity	KGVDDB
1	1q21.1	146,556,821	146,590,236	33,416	cn=3	Gain	NBPF13P								No
1	1q21.1	146,698,906	146,724,923	26,018	cn=3	Gain	CHD1L								No
1	1q21.1	146,767,908	146,946,857	178,950	cn=3	Gain	LINC00624								No
1	1q21.2	147,766,567	147,823,816	57,250	cn=3	Gain	MIRS087								No
1	1q21.2	150,082,673	150,865,007	782,335	cn=3	Gain	ADAMTSL4,ADAMTSL4-AS1,ANP32E,APH1A,ARNT,C1orf54,CA14,CIART,CITSK,CITSE,ECM1,ENSA,FALE,C,GOLPH3L,HORMAD1,MCL1,MIR4257,MIR6878,MRPS21,PLEKHO1,PRPF3,RP	PRPF3, CTSK, ECM1, ARNT, ADAMTSL4	Retinitis pigmentosa 18(601414); Pycnodysostosis (265800); Urbach-Wiethe disease (247100); Ectopia lentis		ARNT, MCL1		CTSS	Yes	
1	1q21.3	151,340,680	151,408,434	67,755	cn=3	Gain	POGZ,PSMB4,SELENBP1								No
7	7q36.1	149,510,936	149,585,639	74,704	cn=3	Gain	ATP6V0E2,ATP6V0E2-AS1,SSPO,ZNF862								No
7	7q36.2	153,533,128	153,634,958	101,831	cn=3	Gain	DPP6	DPP6	Mental retardation (616311), Ventricular						Yes
10	10p13	12,809,031	12,894,155	85,125	cn=3	Gain	CAMK1D								No
15	15q11.2	22,342,453	22,548,420	205,968	cn=1	Loss	LOC101927079,LOC727924,MIR1268A,OR4M2,OR4N3P,OR4N4,REREP3								No