

Copy number variation (CNV) analysis

Document No.	2017-10
Issue date	2018-10-15

Stem cell Information

Stem cell line	CMC-hiPSC-005	Institute	KSCR
Cell type	hiPSC	Inspection date	
Banking status	PCB	Issue date	2018-10-15
Passage	p20		
Note	cultured on vitronectin		

Experiment type

SNP chip

Platform	Affymetrix CytoscanHD	Analysis program	Chas 3.1
Reference	hg19	Analysis document	SOP#26-Ver.4

Statistics

	Total	Gain	Loss	
the number of total CNVs	9			
the number of manually filtered CNVs	1	0	1	
the number of CNVs excluded Korean normal CNV DB (KGVDB)	0	0	0	

Result of Data Analysis

List of CNVs

	Total	Gain	Loss	Cytoband
The number of total CNV calls	1	0	1	
The number of Pathogenic CNVs	1	0	1	4q35.1
The number of Recurrent CNVs	.	.	.	
The number of stemness-related CNVs	1	0	1	4q35.1
The number of Differentiation-related CNVs	.	.	.	
The number of cancer-related CNVs	1	0	1	4q35.1
The number of immunogenicity-related CNVs	1	0	1	4q35.1

*Recurrent CNVs include CNV gain on 1q41, 12p13.31, 17q25.2 and 20q11.21, CNV loss on 10p11.22

Interpretation

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Stem cell Information			
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Result of Data Analysis

List of CNVs

chr	Cytoband	chr_start	chr_end	length(kbp)	Copy Number	CNV	Genes	Pathogenic CNV	Phenotype	Recurrent CNV	Cancer-related	Stemness-related	Differentiation-related	immunogenicity-related	KGVDB
4	q5.1-35.2	183,995,032	190,957,473	6962.4	1	Loss	WWC2-AS2, WWC2, WWC2-AS1, CLDN22, CLDN24, CDKN2AIP, LOC389247, ING2, RWDD4, TRAPPC11, STOX2, ENPP6, LOC728175, LOC102723766, IRF2, LVCAT8, CASP3, PRIMPOL, CENPU, ACSL1, SLED1, MIR3945HG, MIR3945, LINC01093, MIR455, HELT, SLC25A4, CFAP97, SNX25, LRP2BP, ANKRD37, UFSP2, C4orf47, CCDC110, LOC105377590, PDLIM3, SORBS2, TLR3, FAM149A, FLJ38576, CYP4V2, KLKB1, F11, F11-AS1, MTNR1A, FAT1, LOC339975, LOC100506272, ZFP42,	CDKN2AIP, ING2, TRAPPC11, ENPP6, IRF2, CASP3, PRIMPOL, CENPU, ACSL1, SLC25A4, CFAP97, UFSP2, CCDC110, PDLIM3, SORBS2, TLR3, CYP4V2, KLKB1, F11, MTNR1A, FAT1, ZFP42, FRG1, FRG2	CDKN2AIP (615914), ING2 (604215), TRAPPC11 (614138), ENPP6 (616983), IRF2 (147576), CASP3 (600636), PRIMPOL (615421), CENPU (611511), AC SL1 (152425), SLC25A4 (103220), CFA P97 (616047), UFS P2 (611482), CCDC110 (609488), PDLIM3 (605889), SORBS2 (616349), TLR3 (603029), CYP4V2 (608614), KLKB1 (229000), F11 (264900), MTNR1A (600665), FAT1 (600976), ZFP42 (614572), FRG1		IRF2, FAT1	ZFP42		IRF2, CASP3, TLR3	Yes