

Stem cell Information			
Stem cell line	CHA-hES15	Institute	KSCR
Cell type	hESC	Banking year	2016
Banking status	DCB		
Passage	p28		
Single Nucleotide Variation(SNV), short Insert/Deletion(INDEL) analysis			
Experiment of data Analysis			
Whole Exome Sequencing			
Instrument	Illumina HiSeq2500	Analysis program	GATK (UnifiedCaller, v2.3)
Exome Kit	Agilent SureSelectXT V5+UTR	Analysis document	SOP-BI-01
Read length (bp)	101x2	Reference	hg19
Statistics			
Total reads		149,706,296	
Total yield (Gbp)		15.1	
Throughput mean depth(X)		202	
On-target reads		130,279,595	
On-target depth(X)		117	
Result of data Analysis			
SNV, INDEL call			
		Stemness	Differentiation
Total variant		75,973	
SNV		68,095	
DEL		4,018	
INS		3,860	
Coding regions		20,907	
Inframeshift		191	11
Frameshift		162	17
Missense		9,962	0
Nonsense		77	0
Synonymous		10,515	0

* Variant calling condition: GATK, DP(>=10)+MQ(>=58)+AF(>=0.3)

Stemness gene

	Chr	Chr_start	dbSNP135_full	Ref	Alt	Type	Hom_het	Change	Gene	HGVS_C	HGVS_P	비고
1	chr1	12175729	rs1763642	C	T	SNP	Homozygous	missense_variant	TNFRSF8	c.889C>T	p.Arg297Cys	NM_001243.4
2	chr1	12186058	rs2230625	A	G	SNP	Heterozygous	missense_variant	TNFRSF8	c.1204A>G	p.Ser402Gly	NM_001243.4
3	chr1	21894735	rs3200254	T	C	SNP	Heterozygous	missense_variant	ALPL	c.787T>C	p.Tyr263His	NM_000478.4
4	chr1	226076669	rs41310561	C	T	SNP	Heterozygous	missense_variant	LEFTY1	c.98G>A	p.Arg33Gln	NM_020997.3
5	chr3	109052732	rs3762648	T	C	SNP	Heterozygous	missense_variant	DPPA4	c.163A>G	p.Ile55Val	NM_018189.3
6	chr4	89061114	rs2231137	C	T	SNP	Heterozygous	missense_variant	ABCG2	c.34G>A	p.Val12Met	NM_004827.2
7	chr5	98192164	rs138635992	AAGG	A	DEL	Heterozygous	inframe_deletion	CHD1	c.5050_5052delC	p.Pro1684del	NM_001270.2
8	chr7	131195712	rs12670788	G	A	SNP	Heterozygous	missense_variant	PODXL	c.581C>T	p.Ser194Leu	NM_001018111.2
9	chr7	131195959	rs3735035	C	T	SNP	Heterozygous	missense_variant	PODXL	c.334G>A	p.Gly112Ser	NM_001018111.2
10	chr12	7842932	rs1819884;rs386526	C	T	SNP	Heterozygous	missense_variant	GDF3	c.637G>A	p.Gly213Arg	NM_020634.1
11	chr13	28537317	rs1805107	G	A	SNP	Heterozygous	missense_variant	CDX2	c.877C>T	p.Pro293Ser	NM_001265.4

Differentiation gene

	Chr	Chr_start	dbSNP135_full	Ref	Alt	Type	Hom_het	Change	Gene	HGVS_C	HGVS_P	비고
1	chr1	94544234	rs3112831	T	C	SNP	Heterozygous	missense_variant	ABCA4	c.1268A>G	p.His423Arg	NM_000350.2
2	chr1	226076669	rs41310561	C	T	SNP	Heterozygous	missense_variant	LEFTY1	c.98G>A	p.Arg33Gln	NM_020997.3
3	chr2	171225841	rs4668246	A	G	SNP	Homozygous	missense_variant	MYO3B	c.925A>G	p.Lys309Glu	NM_138995.4
4	chr2	171260787	rs6736609	G	A	SNP	Heterozygous	missense_variant	MYO3B	c.2308G>A	p.Val770Ile	NM_138995.4
5	chr2	171356274	rs10185178	G	A	SNP	Homozygous	missense_variant	MYO3B	c.3245G>A	p.Arg1082Lys	NM_138995.4
6	chr2	234863788	rs7593557	G	A	SNP	Heterozygous	missense_variant	TRPM8	c.1256G>A	p.Ser419Asn	NM_024080.4
7	chr3	27763427	2715125;rs18741	G	C	SNP	Heterozygous	missense_variant	EOMES	c.359C>G	p.Ala120Gly	NM_001278182.1
8	chr3	27763427	rs74198;rs368178	G	GCGGCGC	INS	Homozygous	inframe_insertion	EOMES	c.58_359insGCGG	p.119_Ala120insG	NM_001278182.1
9	chr4	55139771	rs35597368	T	C	SNP	Heterozygous	missense_variant	PDGFRA	c.1432T>C	p.Ser478Pro	NM_006206.4
10	chr4	57516896	rs4371677	G	A	SNP	Heterozygous	missense_variant	HOPX	c.281C>T	p.Pro94Leu	NM_001145460.1
11	chr8	97614625	rs3816208	G	A	SNP	Heterozygous	missense_variant&splice_region_variant	SDC2	c.175G>A	p.Ala59Thr	NM_002998.3
12	chr10	72195439	rs1904589	T	C	SNP	Homozygous	missense_variant	NODAL	c.494A>G	p.His165Arg	NM_018055.4
13	chr13	28537317	rs1805107	G	A	SNP	Heterozygous	missense_variant	CDX2	c.877C>T	p.Pro293Ser	NM_001265.4
14	chr14	73711394	rs2280792	A	G	SNP	Heterozygous	missense_variant	PAPLN	c.97A>G	p.Ser33Gly	NM_173462.3
15	chr14	73727509	rs177389	T	G	SNP	Heterozygous	missense_variant	PAPLN	c.1997T>G	p.Met666Arg	NM_173462.3
16	chr14	73729069	rs148072678	C	T	SNP	Heterozygous	missense_variant	PAPLN	c.2176C>T	p.Arg726Trp	NM_173462.3
17	chr16	66432423	rs49970;rs386513	T	C	SNP	Homozygous	missense_variant	CDH5	c.1550T>C	p.Ile517Thr	NM_001795.4