

Stem cell Information			
Stem cell line	hFmiPS1	Institute	KSCR
Cell type	hiPSC	Banking year	2014
Banking status	DCB		
Passage	p30		
Single Nucleotide Variation(SNV), short Insert/Deletion(INDEL) analysis			
Experiment of data Analysis			
Whole Exome Sequencing			
Instrument	Illumina HiSeq2500	Analysis program	GATK (UnifiedGenotyper, v2.3)
Exome Kit	Agilent SureSelectXT V4+UTR	Analysis document	SOP-BI-01
Read length (bp)	101x2	Reference	hg19
Statistics			
Total reads		223,683,782	
Total yield (Gbp)		22.4	
Throughput mean depth(X)		298	
On-target reads		163,196,121	
On-target depth(X)		195	
Result of data Analysis			
SNV, INDEL call			
		Stemness	Differentiation
Total variant		45,035	
SNV		41,881	
DEL		1,588	
INS		1,566	
Coding regions		10,255	16
Inframeshift		192	1
Frameshift		176	0
Missense		9,623	15
Nonsense		94	0
Synonymous		10,245	
* 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	21894735	rs3200254	T	C	SNP	Heterozygous	missense_variant	ALPL	c.787T>C	p.Tyr263His	NM_000478.4
2	chr1	226125385	rs2295418	G	A	SNP	Heterozygous	missense_variant	LEFTY2	c.857C>T	p.Pro286Leu	NM_003240.3
3	chr1	12175729	rs1763642	C	T	SNP	Homozygous	missense_variant	TNFRSF8	c.889C>T	p.Arg297Cys	NM_001243.4
4	chr13	28537317	rs1805107	G	A	SNP	Homozygous	missense_variant	CDX2	c.877C>T	p.Pro293Ser	NM_001265.4
5	chr3	109052732	rs3762648	T	C	SNP	Homozygous	missense_variant	DPPA4	c.163A>G	p.Ile55Val	NM_018189.3
6	chr4	89020503	rs192169063	A	G	SNP	Heterozygous	missense_variant	ABCG2	c.1465T>C	p.Phe489Leu	NM_004827.2
7	chr4	89052323	rs2231142	G	T	SNP	Heterozygous	missense_variant	ABCG2	c.421C>A	p.Gln141Lys	NM_004827.2
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	chr8	128752696	.	A	T	SNP	Heterozygous	missense_variant	MYC	c.857A>T	p.Gln286Leu	NM_002467.4

**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	226125385	rs2295418	G	A	SNP	Heterozygous	missense_variant	LEFTY2	c.857C>T	p.Pro286Leu	NM_003240.3
3	chr10	72195439	rs1904589	T	C	SNP	Homozygous	missense_variant	NODAL	c.494A>G	p.His165Arg	NM_018055.4
4	chr12	48367976	rs2070739	C	T	SNP	Heterozygous	missense_variant	COL2A1	c.4213G>A	p.Gly1405Ser	NM_001844.4
5	chr12	48368541	rs721427;rs386525	C	T	SNP	Heterozygous	missense_variant	COL2A1	c.3991G>A	p.Val1331Ile	NM_001844.4
6	chr13	28537317	rs1805107	G	A	SNP	Homozygous	missense_variant	CDX2	c.877C>T	p.Pro293Ser	NM_001265.4
7	chr14	73711394	rs2280792	A	G	SNP	Heterozygous	missense_variant	PAPLN	c.97A>G	p.Ser33Gly	NM_173462.3
8	chr14	73727509	rs177389	T	G	SNP	Homozygous	missense_variant	PAPLN	c.1997T>G	p.Met666Arg	NM_173462.3
9	chr14	73727926	rs2242616	G	C	SNP	Heterozygous	missense_variant	PAPLN	c.2088G>C	p.Gln696His	NM_173462.3
10	chr14	73733285	rs61745771	G	A	SNP	Heterozygous	missense_variant	PAPLN	c.3259G>A	p.Ala1087Thr	NM_173462.3
11	chr14	73735366	rs4903104	C	T	SNP	Heterozygous	missense_variant	PAPLN	c.3521C>T	p.Thr1174Met	NM_173462.3
12	chr16	66432423	rs49970;rs386513	T	C	SNP	Homozygous	missense_variant	CDH5	c.1550T>C	p.Ile517Thr	NM_001795.4
13	chr2	234863788	rs7593557	G	A	SNP	Heterozygous	missense_variant	TRPM8	c.1256G>A	p.Ser419Asn	NM_024080.4
14	chr3	27763427	rs74198;rs368178	G	GCGGCGC	INS	Homozygous	inframe_insertion	EOMES	c.358_359insG	p.Ala119_Ala12	NM_001278182.1
15	chr4	55139771	rs35597368	T	C	SNP	Heterozygous	missense_variant	PDGFRA	c.1432T>C	p.Ser478Pro	NM_006206.4
16	chr8	97620596	.	C	T	SNP	Heterozygous	missense_variant	SDC2	c.340C>T	p.Leu114Phe	NM_002998.3