

| Stem cell Information   |                             |                   |                 |
|---|-----------------------------|-------------------|-----------------|
| Stem cell line  | CMC-iPSC-009                | Institute         | KSCR            |
| Cell type   | hiPSC                       | Banking year      | 2017            |
| Banking status  | PCB                         |                   |                 |
| Passage   | p26                         |                   |                 |
| Single Nucleotide Variation(SNV), short Insert/Deletion(INDEL) analysis |                             |                   |                 |
| Experiment of data Analysis   |                             |                   |                 |
| Whole Exome Sequencing  |                             |                   |                 |
| Instrument  | Illumina HiSeq2500          | Analysis program  | GATK (v3.5)     |
| Exome Kit   | Agilent SureSelectXT V5+UTR | Analysis document | SOP-BI-01       |
| Read length (bp)  | 101x2                       | Reference         | hg19            |
| Statistics  |                             |                   |                 |
| Total reads   |                             | 202,604,670       |                 |
| Total yield (Gbp)   |                             | 20.40             |                 |
| Throughput mean depth(X)  |                             | 272               |                 |
| On-target reads   |                             | 104,708,302       |                 |
| On-target depth(X)  |                             | 142               |                 |
| Result of data Analysis   |                             |                   |                 |
| SNV, INDEL call   |                             |                   |                 |
|   |                             | Stemness          | Differentiation |
| <b>Total variant</b>  |                             | 102,289           |                 |
|   | <b>SNV</b>                  | 91,626            |                 |
|   | <b>DEL</b>                  | 5,411             |                 |
|   | <b>INS</b>                  | 5,252             |                 |
| <b>Coding regions</b>   |                             | 22,192            | 9 21            |
|   | <b>Synonymous</b>           | 10,312            | - -             |
|   | <b>Missense</b>             | 9,489             | 9 20            |
|   | <b>Inframeshift</b>         | 146               | - -             |
|   | <b>Frameshift</b>           | 150               | - 1             |
|   | <b>Nonsense</b>             | 137               | - -             |
|   | <b>SpliceRegion</b>         | 1,958             | - -             |
| * 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 | 4   | 89061114  | rs2231137     | C   | T   | SNP  | '0/1    | missense_variant | ABCG2   | c.34G>A  | 'p.Val12Met  | NM_001257386.1 |
| 2 | 1   | 21894735  | rs3200254     | T   | C   | SNP  | '1/1    | missense_variant | ALPL    | c.556T>C | 'p.Tyr186His | NM_001177520.1 |
| 3 | 13  | 28537317  | rs1805107     | G   | A   | SNP  | '1/1    | missense_variant | CDX2    | c.877C>T | 'p.Pro293Ser | NM_001265.4    |
| 4 | 3   | 109052732 | rs3762648     | T   | C   | SNP  | '1/1    | missense_variant | DPPA4   | c.163A>G | 'p.Ile55Val  | NM_018189.3    |
| 5 | 2   | 47601106  | rs1126497     | T   | C   | SNP  | '1/1    | missense_variant | EPCAM   | c.344T>C | 'p.Met115Thr | NM_002354.2    |
| 6 | 1   | 63789015  | rs2274188     | G   | T   | SNP  | '0/1    | missense_variant | FOXD3   | c.286G>T | 'p.Val96Leu  | NM_012183.2    |
| 7 | 7   | 131193739 | rs3212298     | C   | T   | SNP  | '1/1    | missense_variant | PODXL   | c.976G>A | 'p.Val326Ile | NM_005397.3    |
| 8 | 1   | 12186058  | rs2230625     | A   | G   | SNP  | '0/1    | missense_variant | TNFRSF8 | c.871A>G | 'p.Ser291Gly | NM_001281430.2 |
| 9 | 1   | 12175729  | rs1763642     | C   | T   | SNP  | '1/1    | missense_variant | TNFRSF8 | c.556C>T | 'p.Arg186Cys |                |

**Differentiation gene**

|    | Chr | Chr_start | dbsnp135_full | Ref | Alt | Type | Hom_het | Change                                 | Gene   | HGVS_C     | HGVS_P        | 비고             |
|----|-----|-----------|---------------|-----|-----|------|---------|--|--------|------------|---------------|----------------|
| 1  | 1   | 94544234  | rs3112831     | T   | C   | SNP  | '0/1    | missense_variant                       | ABCA4  | c.1268A>G  | 'p.His423Arg  | NM_000350.2    |
| 2  | 17  | 4539060   | .             | G   | GA  | INS  | '0/1    | frameshift_variant                     | ALOX15 | c.1154dupT | 'p.Phe386fs   | NM_001140.3    |
| 3  | 18  | 59174759  | rs1943330     | C   | A   | SNP  | '0/1    | missense_variant                       | CDH20  | c.983C>A   | 'p.Pro328His  | NM_031891.2    |
| 4  | 16  | 66432423  | rs1049970     | T   | C   | SNP  | '0/1    | missense_variant                       | CDH5   | c.1550T>C  | 'p.Ile517Thr  | NM_001795.4    |
| 5  | 13  | 28537317  | rs1805107     | G   | A   | SNP  | '1/1    | missense_variant                       | CDX2   | c.877C>T   | 'p.Pro293Ser  | NM_001265.4    |
| 6  | 12  | 48367976  | rs2070739     | C   | T   | SNP  | '0/1    | missense_variant                       | COL2A1 | c.4006G>A  | 'p.Gly1336Ser | NM_033150.2    |
| 7  | 14  | 38061742  | rs7144658     | C   | T   | SNP  | '1/1    | missense_variant                       | FOXA1  | c.247G>A   | 'p.Ala83Thr   | NM_004496.3    |
| 8  | 18  | 19751148  | rs116262672   | G   | C   | SNP  | '0/1    | missense_variant                       | GATA6  | c.43G>C    | 'p.Gly15Arg   | NM_005257.5    |
| 9  | 2   | 171239677 | rs34273653    | A   | G   | SNP  | '0/1    | missense_variant                       | MYO3B  | c.1163A>G  | 'p.Asn388Ser  | NM_001083615.3 |
| 10 | 2   | 171260787 | rs6736609     | G   | A   | SNP  | '0/1    | missense_variant                       | MYO3B  | c.2308G>A  | 'p.Val770Ile  | NM_001083615.3 |
| 11 | 2   | 171260797 | rs33962844    | A   | G   | SNP  | '0/1    | missense_variant                       | MYO3B  | c.2318A>G  | 'p.Glu773Gly  | NM_001083615.3 |
| 12 | 2   | 171356274 | rs10185178    | G   | A   | SNP  | '1/1    | missense_variant                       | MYO3B  | c.3245G>A  | 'p.Arg1082Lys | NM_001083615.3 |
| 13 | 2   | 171225841 | rs4668246     | A   | G   | SNP  | '0/1    | missense_variant                       | MYO3B  | c.925A>G   | 'p.Lys309Glu  | NM_001083615.3 |
| 14 | 10  | 72195439  | rs1904589     | T   | C   | SNP  | '0/1    | missense_variant                       | NODAL  | c.494A>G   | 'p.His165Arg  | NM_018055.4    |
| 15 | 4   | 71068489  | rs3196714     | T   | C   | SNP  | '0/1    | missense_variant                       | ODAM   | c.665T>C   | 'p.Ile222Thr  | NM_017855.3    |
| 16 | 14  | 73727509  | rs177389      | T   | G   | SNP  | '1/1    | missense_variant                       | PAPLN  | c.1997T>G  | 'p.Met666Arg  | NM_173462.3    |
| 17 | 14  | 73711394  | rs2280792     | A   | G   | SNP  | '0/1    | missense_variant                       | PAPLN  | c.97A>G    | 'p.Ser33Gly   | NM_173462.3    |
| 18 | 14  | 73727926  | rs2242616     | G   | C   | SNP  | '0/1    | missense_variant                       | PAPLN  | c.2088G>C  | 'p.Gln696His  | NM_173462.3    |
| 19 | 11  | 31814819  | .             | C   | T   | SNP  | '0/1    | missense_variant                       | PAX6   | c.1199G>A  | 'p.Ser400Asn  | NM_001310159.1 |
| 20 | 8   | 97614625  | rs3816208     | G   | A   | SNP  | '1/1    | missense_variant&splice_region_variant | SDC2   | c.175G>A   | 'p.Ala59Thr   | NM_002998.3    |
| 21 | 8   | 55372118  | .             | A   | T   | SNP  | '0/1    | missense_variant                       | SOX17  | c.808A>T   | 'p.Met270Leu  | NM_022454.3    |