

Figure 6 iPS 細胞及び hMSCs における LINE-1s mRNA の発現

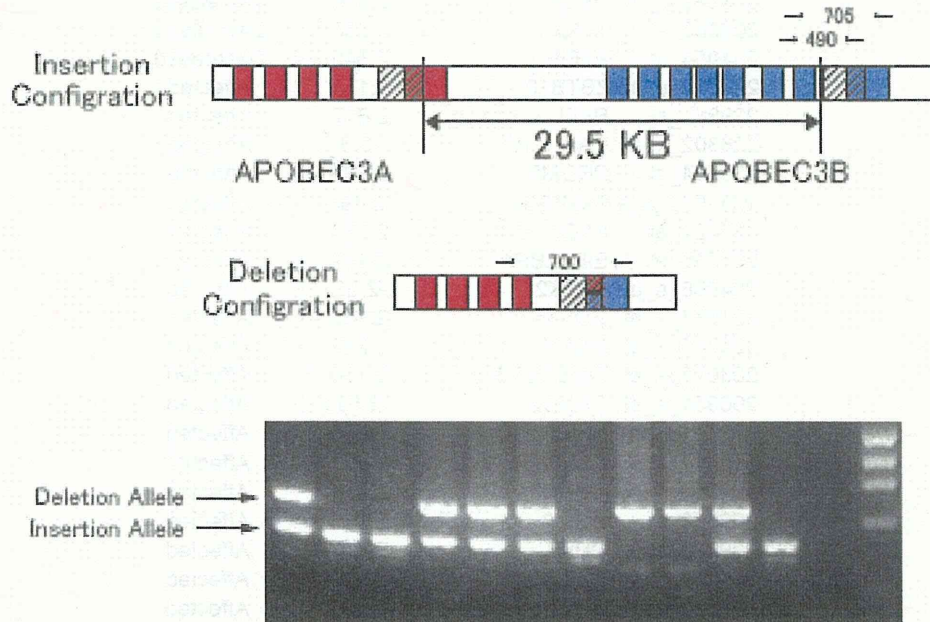


Figure 7 PCR による A3B 遺伝子型解析

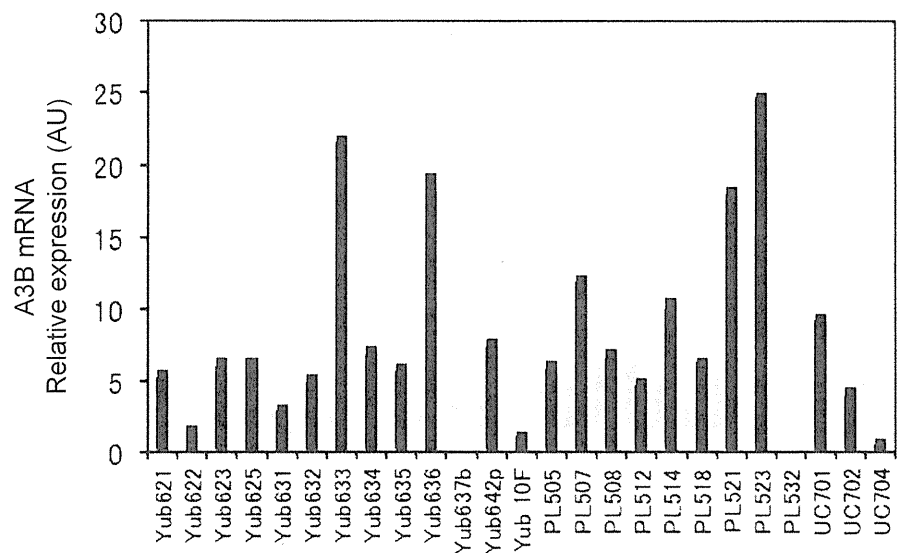


Figure 8 hMSCs における A3B mRNA の発現比較

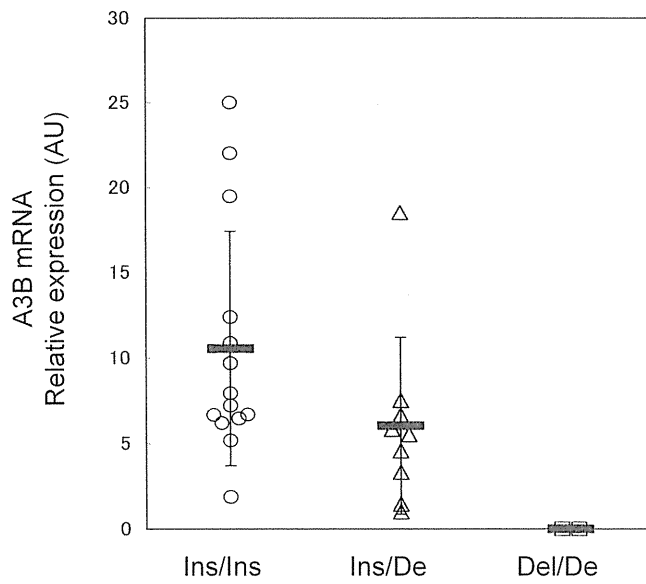


Figure 9 A3B 遺伝子型による A3B mRNA の発現比較

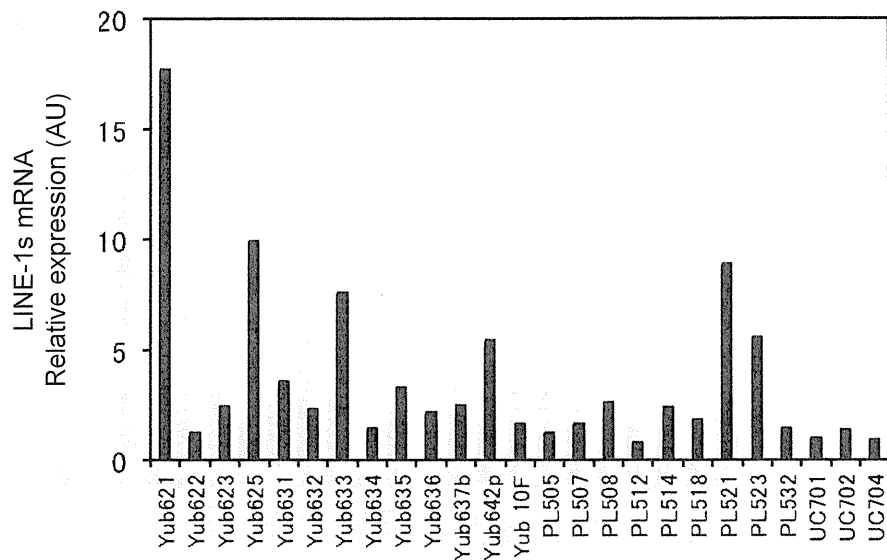


Figure 10 hMSCs における LINE-1s mRNA の発現比較

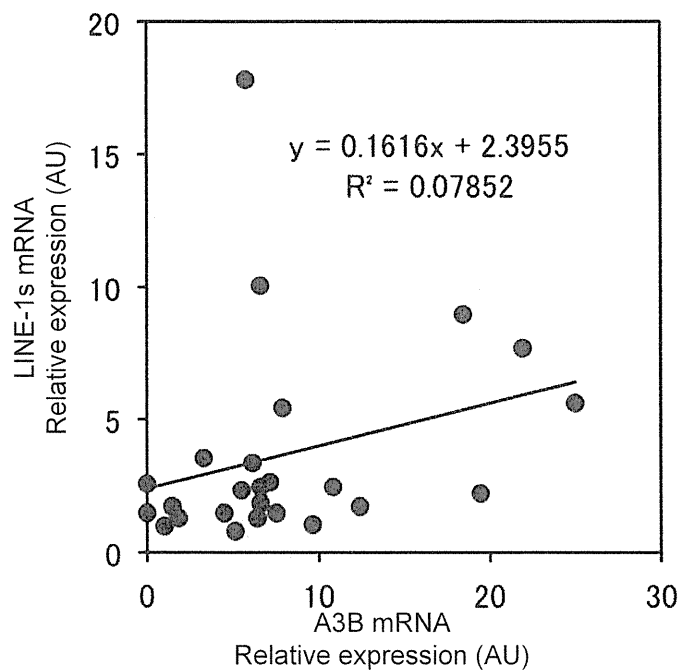


Figure 11 hMSCs における A3B, LINE-1s mRNA 発現の相関

[GENETYX-MAC: Multiple Alignment]
Date 2019.11.19

LINE-1.3(PCR)	1	1
L1 b-thal1(PCR)	1	1
L1 RP(PCR)	1	1
11-8For	1 NNNNNNNNNNNNNNNNNNNNNNNCCGANGTTCGANGGTTTCGATAGGTTT---TATATC---G	54
11-13For	1 ---NNNNNNNNNNNNNNNNNNNNNNCTNNGGTGANGGTTTCGATAGGTTT---TATATC---G	50
11-25For	1 ---NNNNNNNNNNNNNNNNNNNNNNCTGATAGGTTTCGATAGGTTT---TATATC---G	51
11-30For	1 ---NNNNNNNNNNNNNNNNNNNNNNGANGGTTTCGATAGGTTT---TATATC---G	47
11-3Rev	1 NNNNNNNNNNNNNNNNNNNNNNNGNGCGGCCCTCTAGACCTAGTGGGTTCCCGCGGCTCAGG	60
LINE-1.3(PCR)	1 -----CAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	47
L1 b-thal1(PCR)	1 -----CAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	47
L1 RP(PCR)	1 -----CAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	47
11-8For	55 AATTCCTCAATCTCAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	114
11-13For	51 AATTCCTCAATCTCAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	110
11-25For	52 AATTCCTCAATCTCAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	111
11-30For	48 AATTCCTCAATCTCAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	107
11-3Rev	61 AATTCCTCAATCTCAGGGCAATCAGGCGAGGAGAGGAAATAAAGGGTATTCAAATAGGAA	120
LINE-1.3(PCR)	48 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	106
L1 b-thal1(PCR)	48 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	106
L1 RP(PCR)	48 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	106
11-8For	115 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	173
11-13For	115 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	170
11-25For	112 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	170
11-30For	108 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	164
11-3Rev	121 AAGAGGAAGTCAAAATGTCCCTGTTTCGACAGCAGCATGATGTTAT---CTAGAAAACCC	179
LINE-1.3(PCR)	107 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	166
L1 b-thal1(PCR)	107 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	166
L1 RP(PCR)	107 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	166
11-8For	174 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	230
11-13For	171 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	230
11-25For	171 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	230
11-30For	165 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	222
11-3Rev	180 ATGCTTCAGGCCAAAATCTCCTTAAGCTGATAAGCAACTCAGCAAACTTCAGGATAT	239
LINE-1.3(PCR)	167 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	226
L1 b-thal1(PCR)	167 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	226
L1 RP(PCR)	167 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	226
11-8For	234 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	293
11-13For	231 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	290
11-25For	231 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	282
11-30For	223 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	282
11-3Rev	240 AAAATCAATGTACAAAATACAAAGCAATCTTATACACCAACCAACAGCAAAACAGAGAG	299
LINE-1.3(PCR)	227 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	286
L1 b-thal1(PCR)	227 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	286
L1 RP(PCR)	227 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	286
11-8For	297 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	353
11-13For	291 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	350
11-25For	283 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	342
11-30For	283 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	342
11-3Rev	300 CAAATCATGGTGAACCTCCATTCGAAATGCTTCAAGAGAAATAAATACTAGGAATC	359
LINE-1.3(PCR)	287 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	346
L1 b-thal1(PCR)	287 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	346
L1 RP(PCR)	287 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	346
11-8For	354 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	413
11-13For	351 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	410
11-25For	343 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	402
11-30For	343 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	402
11-3Rev	360 CAACCTCAAGGGATGTGAAGGACCTCTTCAAGGAGAACTCAAAACCACTGCTCAAGAA	418
LINE-1.3(PCR)	347 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	406
L1 b-thal1(PCR)	347 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	406

L1 RP(PCR)	347 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	406
11-8For	424 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	471
11-13For	411 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	467
11-25For	403 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	462
11-30For	403 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	462
11-3Rev	420 ATAAAAGAGGATCAAAACAATGGAAGAACATTCATGCTCATGGTAGGAAGATCAAT	470
LINE-1.3(PCR)	407 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	466
L1 b-thal1(PCR)	407 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	466
L1 RP(PCR)	407 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	466
11-8For	472 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	531
11-13For	471 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	530
11-25For	463 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	522
11-30For	463 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	522
11-3Rev	480 ATCTGAAAAATGGCCATCTGCCCAAGGTAATTTAGATTCAATGCCATCCCATCAAG	539
LINE-1.3(PCR)	467 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	525
L1 b-thal1(PCR)	467 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	525
L1 RP(PCR)	467 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	525
11-8For	532 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	591
11-13For	531 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	589
11-25For	523 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	581
11-30For	523 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	581
11-3Rev	540 ATCTCAATGACTTCTTCACAGAAATGGAAAAA---TACTTTAAAGTTCATATGGAAACA	598
LINE-1.3(PCR)	526 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	585
L1 b-thal1(PCR)	526 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	585
L1 RP(PCR)	526 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	585
11-8For	592 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	651
11-13For	590 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	649
11-25For	582 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	641
11-30For	582 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	641
11-3Rev	599 AAAAAGAGCCCGCATTCGCAAGTCAATCTAAGCCAAAAGAACAAAGCTGGAGGCATCA	658
LINE-1.3(PCR)	586 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	645
L1 b-thal1(PCR)	586 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	645
L1 RP(PCR)	586 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	645
11-8For	645 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	711
11-13For	650 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	700
11-25For	642 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	701
11-30For	642 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	701
11-3Rev	659 ATCTCTGACTTCAAACTATACTACAAGGCTACAGTAACCAAACAGCATGGTACTGGTA	718
LINE-1.3(PCR)	646 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	705
L1 b-thal1(PCR)	646 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	705
L1 RP(PCR)	646 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	705
11-8For	712 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	771
11-13For	710 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	769
11-25For	702 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	761
11-30For	702 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	761
11-3Rev	719 CCAAAAACAGAGATATAGATCAATGGAAACAGACAGAGCCCTCAGAAATAATGCCGATAT	775
LINE-1.3(PCR)	706 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	765
L1 b-thal1(PCR)	706 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	765
L1 RP(PCR)	706 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	765
11-8For	772 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	831
11-13For	770 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	829
11-25For	762 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	821
11-30For	762 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	820
11-3Rev	776 CTACAACATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATTCCTC	835
LINE-1.3(PCR)	766 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	824
L1 b-thal1(PCR)	766 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	824
L1 RP(PCR)	766 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	824
11-8For	852 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	889
11-13For	830 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	885
11-25For	822 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	880
11-30For	821 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	880
11-3Rev	836 ATTTAAATAATGGTCTGGGAAACCTGGTAGCCATA---TGTAGAAAGCTGAAATGGATC	894
LINE-1.3(PCR)	825 CCTCTCTACACCTTATACAAAATCAATTCAGATGGATTAAGATTTAAAGCTTAAAC	884
L1 b-thal1(PCR)	825 CCTCTCTACACCTTATACAAAATCAATTCAGATGGATTAAGATTTAAAGCTTAAAC	884

Figure 12A PL523 (Ins/Ins) における LINE-1s mRNA の sequence

{GENETYX-MAC: Multiple Alignment}
Date 2011.11.29

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LINE-1.3(PCR) 1 ----- 1  
L1 b-thal(PCR) 1 ----- 1  
L1 RP(PCR) 1 ----- 1  
1-1For 1 NNNNNNNNNNNNNNNNNNGGCTCGANGGATCGATAAGCTTGATCGAATGCCAAT 60  
1-4For 1 NNNNNNNNNNNNNNNNNCGGCTCGANGGATCGATAAGCTTGATCGAATGCCAAT 60  
1-5For 1 NNNNNNNNNNNNNNNNNCGGCTCGANGGATCGATAAGCTTGATCGAATGCCAAT 59  
1-6For 1 NNNNNNNNNNNNNNNNNCGGCTCGANGGATCGATAAGCTTGATCGAATGCCAAT 58  
1-8For 1 NNNNNNNNNNNNNNNNNCGGCTCGANGGATCGATAAGCTTGATCGAATGCCAAT 57  
LINE-1.3(PCR) 1 ---CAGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 57  
L1 b-thal(PCR) 1 ---CAGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 57  
L1 RP(PCR) 1 ---CAGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 57  
1-1For 61 ACT---AGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 119  
1-4For 61 ACT---AGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 119  
1-5For 60 ACT---AGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 119  
1-6For 59 ACT---AGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 116  
1-8For 60 ACT---AGGGCAATCAGGCAGGAGAAAGAAATAAGGGTATTCATATTAGGAAGAAGGAAGT 118  
LINE-1.3(PCR) 58 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 117  
L1 b-thal(PCR) 58 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 117  
L1 RP(PCR) 58 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 117  
1-1For 120 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 179  
1-4For 120 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 179  
1-5For 128 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 179  
1-6For 117 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 176  
1-8For 119 CAATTTGCCCTGTTTGCAGTGCATGATGTTATCTAGAAAACCCCATGCTCCAGG 178  
LINE-1.3(PCR) 118 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 177  
L1 b-thal(PCR) 118 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 177  
L1 RP(PCR) 118 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 177  
1-1For 180 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 239  
1-4For 180 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 239  
1-5For 180 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 239  
1-6For 177 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 236  
1-8For 179 CCAAAATCTCCTTAAGCTGATAAGCAACTTCAGCAAGTCTCAGGATACAAAATCAATG 238  
LINE-1.3(PCR) 178 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 237  
L1 b-thal(PCR) 178 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 237  
L1 RP(PCR) 178 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 237  
1-1For 240 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 299  
1-4For 240 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 299  
1-5For 240 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 299  
1-6For 237 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 296  
1-8For 239 CCAAAAATCACAAAGCTTCTTATACACCAATACAGACAACACAGAGAGCCAAATCATGG 298  
LINE-1.3(PCR) 238 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 297  
L1 b-thal(PCR) 238 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 297  
L1 RP(PCR) 238 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 297  
1-1For 300 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 359  
1-4For 300 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 359  
1-5For 300 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 359  
1-6For 297 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 356  
1-8For 299 TGAATCCCATTTCTAATGCTTCAAGAGAAATAAATACTAGGAATCCAATCTACAAG 358  
LINE-1.3(PCR) 298 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 353  
L1 b-thal(PCR) 298 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 353  
L1 RP(PCR) 298 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 353  
1-1For 360 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 415  
1-4For 360 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 415  
1-5For 360 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 415  
1-6For 357 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 416  
1-8For 359 GGTGTGAAGGACCTCTTCAAGGAACTACA---CACCTGCTCAAGGAAATAAAG 414  
LINE-1.3(PCR) 354 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 413  
L1 b-thal(PCR) 354 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 413
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L1 RP(PCR) 354 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 413  
1-1For 416 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 472  
1-4For 416 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 475  
1-5For 416 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 476  
1-6For 417 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 476  
1-8For 415 AGGACACAAACAAATGGGAAGCAATTCATGCTCATGGTAGGAAGTAATCAATCTCGTGA 474  
LINE-1.3(PCR) 414 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 473  
L1 b-thal(PCR) 414 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 473  
L1 RP(PCR) 414 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 473  
1-1For 417 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 532  
1-4For 417 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 532  
1-5For 417 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 535  
1-6For 417 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 536  
1-8For 417 AAATGGCCATCTGCCCAAGGTAATTTACAGATTCAATGCCATCCCATCAAGCTACCAA 534  
LINE-1.3(PCR) 474 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 533  
L1 b-thal(PCR) 474 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 533  
L1 RP(PCR) 474 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 533  
1-1For 533 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 592  
1-4For 536 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 595  
1-5For 536 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 595  
1-6For 537 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 591  
1-8For 535 TGACTTCTTCCAGAAATGGAAAAAATCTTTAAAGTTTCATATGGAACCAAAAAGAG 594  
LINE-1.3(PCR) 534 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 593  
L1 b-thal(PCR) 534 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 593  
L1 RP(PCR) 534 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 593  
1-1For 593 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 652  
1-4For 596 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 655  
1-5For 596 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 655  
1-6For 592 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 651  
1-8For 595 CCGCATTTGCCAAGTCAATCTTAAGCAAAAAGAAACAAAGCTGGAGGATCACACTCTG 654  
LINE-1.3(PCR) 594 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 653  
L1 b-thal(PCR) 594 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 653  
L1 RP(PCR) 594 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 653  
1-1For 653 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 712  
1-4For 656 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 715  
1-5For 656 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 715  
1-6For 652 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 714  
1-8For 655 ACTTCAAATATACTACAAGGCTACAGTAACCAAAACAGCATGGTACTGGTACCAAAAC 711  
LINE-1.3(PCR) 654 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 711  
L1 b-thal(PCR) 654 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 711  
L1 RP(PCR) 654 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 711  
1-1For 713 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 770  
1-4For 716 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 775  
1-5For 716 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 773  
1-6For 715 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 769  
1-8For 715 GAGATATAGATCAATGGACAGAACAGAGCCCTCAGAAATAATGCCGATA---TCTACAA 772  
LINE-1.3(PCR) 712 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 770  
L1 b-thal(PCR) 712 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 770  
L1 RP(PCR) 712 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 770  
1-1For 771 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 829  
1-4For 776 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 834  
1-5For 774 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 832  
1-6For 770 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 829  
1-8For 773 TATCTGATCTTTGACAAACCTGAGAAAAACAGCAATGGGAAAGGATCCCTATT---A 831  
LINE-1.3(PCR) 771 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 830  
L1 b-thal(PCR) 771 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 830  
L1 RP(PCR) 771 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 830  
1-1For 830 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 889  
1-4For 835 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 894  
1-5For 833 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 892  
1-6For 830 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 891  
1-8For 832 ATAAATGGTCTGGGAAACCTGGCTAGCCATATGTAAGAAAGCTGAACTGGATCCCTCTC 889  
LINE-1.3(PCR) 831 TTACACTTATACAAAATCAATTCAGGATGGATTAAGTTTAAAGTTTAACTTAAAC 890  
L1 b-thal(PCR) 831 TTACACTTATACAAAATCAATTCAGGATGGATTAAGTTTAAAGTTTAACTTAAAC 890
```

Figure 12B Yub621 (Ins/Del) における LINE-1s mRNA の sequence

[GENETEX-MAG: Multiple Alignment]
Date: 2013.11.19

```
LINE-1.3(PCR) 1 ----- 1  
L1 b-thal(PCR) 1 ----- 1  
L1 RP(PCR) 1 ----- 1  
3-1 For 1 ----- 1  
3-6 For 1 ----- 1  
3-10 For 1 ----- 1  
3-13 For 1 ----- 1  
3-2 Rev 1 ----- 1  
LINE-1.3(PCR) 1 ----- 50  
L1 b-thal(PCR) 1 ----- 50  
L1 RP(PCR) 1 ----- 50  
3-1 For 1 ----- 50  
3-6 For 34 ----- 113  
3-10 For 49 ----- 188  
3-13 For 52 ----- 121  
3-2 Rev 61 ----- 170  
LINE-1.3(PCR) 51 ----- 110  
L1 b-thal(PCR) 51 ----- 110  
L1 RP(PCR) 51 ----- 110  
3-1 For 111 ----- 170  
3-6 For 114 ----- 173  
3-10 For 109 ----- 168  
3-13 For 112 ----- 171  
3-2 Rev 123 ----- 180  
LINE-1.3(PCR) 111 ----- 179  
L1 b-thal(PCR) 111 ----- 179  
L1 RP(PCR) 111 ----- 179  
3-1 For 174 ----- 230  
3-6 For 174 ----- 233  
3-10 For 169 ----- 228  
3-13 For 172 ----- 231  
3-2 Rev 181 ----- 240  
LINE-1.3(PCR) 171 ----- 230  
L1 b-thal(PCR) 171 ----- 230  
L1 RP(PCR) 171 ----- 230  
3-1 For 231 ----- 290  
3-6 For 234 ----- 293  
3-10 For 229 ----- 288  
3-13 For 242 ----- 301  
3-2 Rev 241 ----- 290  
LINE-1.3(PCR) 231 ----- 290  
L1 b-thal(PCR) 231 ----- 290  
L1 RP(PCR) 231 ----- 290  
3-1 For 291 ----- 350  
3-6 For 294 ----- 353  
3-10 For 289 ----- 348  
3-13 For 292 ----- 351  
3-2 Rev 301 ----- 360  
LINE-1.3(PCR) 291 ----- 350  
L1 b-thal(PCR) 291 ----- 350  
L1 RP(PCR) 291 ----- 350  
3-1 For 351 ----- 410  
3-6 For 354 ----- 413  
3-10 For 349 ----- 408  
3-13 For 352 ----- 411  
3-2 Rev 361 ----- 420  
LINE-1.3(PCR) 351 ----- 410  
L1 b-thal(PCR) 351 ----- 410
```

```
L1 RP(PCR) 351 ----- 410  
3-1 For 411 ----- 470  
3-6 For 414 ----- 473  
3-10 For 409 ----- 468  
3-13 For 412 ----- 471  
3-2 Rev 421 ----- 480  
LINE-1.3(PCR) 411 ----- 470  
L1 b-thal(PCR) 411 ----- 470  
L1 RP(PCR) 411 ----- 470  
3-1 For 471 ----- 530  
3-6 For 474 ----- 533  
3-10 For 469 ----- 528  
3-13 For 472 ----- 531  
3-2 Rev 481 ----- 540  
LINE-1.3(PCR) 471 ----- 530  
L1 b-thal(PCR) 471 ----- 530  
L1 RP(PCR) 471 ----- 530  
3-1 For 531 ----- 590  
3-6 For 534 ----- 593  
3-10 For 529 ----- 588  
3-13 For 532 ----- 591  
3-2 Rev 541 ----- 600  
LINE-1.3(PCR) 531 ----- 589  
L1 b-thal(PCR) 531 ----- 589  
L1 RP(PCR) 531 ----- 589  
3-1 For 594 ----- 650  
3-6 For 589 ----- 647  
3-10 For 592 ----- 650  
3-13 For 601 ----- 659  
LINE-1.3(PCR) 590 ----- 649  
L1 b-thal(PCR) 590 ----- 649  
L1 RP(PCR) 590 ----- 649  
3-1 For 649 ----- 709  
3-6 For 652 ----- 712  
3-10 For 648 ----- 707  
3-13 For 651 ----- 710  
3-2 Rev 660 ----- 719  
LINE-1.3(PCR) 650 ----- 709  
L1 b-thal(PCR) 650 ----- 709  
L1 RP(PCR) 650 ----- 709  
3-1 For 709 ----- 769  
3-6 For 711 ----- 770  
3-10 For 713 ----- 772  
3-13 For 708 ----- 767  
3-2 Rev 720 ----- 779  
LINE-1.3(PCR) 710 ----- 769  
L1 b-thal(PCR) 710 ----- 769  
L1 RP(PCR) 710 ----- 769  
3-1 For 769 ----- 829  
3-6 For 771 ----- 830  
3-10 For 773 ----- 832  
3-13 For 772 ----- 831  
3-2 Rev 780 ----- 839  
LINE-1.3(PCR) 770 ----- 829  
L1 b-thal(PCR) 770 ----- 829  
L1 RP(PCR) 770 ----- 829  
3-1 For 829 ----- 889  
3-6 For 831 ----- 890  
3-10 For 833 ----- 892  
3-13 For 828 ----- 887  
3-2 Rev 840 ----- 899  
LINE-1.3(PCR) 830 ----- 889
```

Figure 12C Yub637b (Del/Del) における LINE-1s mRNA の sequence

		To			
		A	C	G	T
From	A		17	43	11
	C	16		20	65
	G	57	17		18
	T	16	51	5	n=10

336 mutations 9087 bp

		To			
		A	C	G	T
From	A		27	77	21
	C	31		26	108
	G	97	31		25
	T	32	65	8	n=13

548 mutations 11752 bp

Figure 13A hMSCs (Ins/Ins) における LINE-1s mRNA の mutation 解析

		To			
		A	C	G	T
From	A		16	40	5
	C	12		9	58
	G	70	20		13
	T	15	49	8	n=10

315 mutations 9,049 bp

		To			
		A	C	G	T
From	A		16	51	19
	C	26		6	66
	G	49	18		19
	T	21	32	11	n=10

334 mutations 8,935 bp

		To			
		A	C	G	T
From	A		8	44	12
	C	21		12	63
	G	60	22		11
	T	12	49	4	n=11

318 mutations 9,958 bp

Figure 13B hMSCs (Ins/Del) における LINE-1s mRNA の mutation 解析

		To			
		A	C	G	T
From	A		3	15	4
	C	5		5	17
	G	25	9		8
	T	5	35	3	n=17
	134 mutations				15428

		To			
		A	C	G	T
From	A		1	4	1
	C	3		2	16
	G	19	2		0
	T	2	17	1	n=11
	68 mutations				9982

Figure 13C hMSCs (Del/Del) における LINE-1s mRNA の mutation 解析

Table 5 日本人由来 hMSCs における A3B 遺伝子型の解析

A3B genotype		A3B genotype		A3B genotype	
Yub621	Ins/Del	PL505	Ins/Ins	UC701	Ins/Ins
Yub622	Ins/Ins	PL507	Ins/Ins	UC702	Ins/Del
Yub623	Ins/Ins	PL508	Ins/Ins	UC704	Ins/Del
Yub625	Ins/Del	PL518	Ins/Ins		
Yub631	Ins/Del	PL512	Ins/Ins		
Yub632	Ins/Del	PL514	Ins/Ins		
Yub633	Ins/Ins	PL521	Ins/Del		
Yub634	Ins/Del	PL523	Ins/Ins		
Yub635	Ins/Ins	PL532	Del/Del		
Yub636	Ins/Ins				
Yub637b	Del/Del				
Yub642p	Ins/Ins				
Yub 10F	Ins/Del				

Genotype (No. of human)

	Ins/Ins	Ins/Del	Del/Del
Yub	6	6	1
PL	7	1	1
UC	1	2	0
total	14	9	2

Allele frequency (%)

Ins	Del
74	26

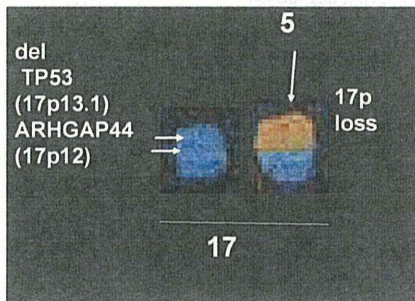
Table 6 HL60 細胞におけるコピー数変化領域の詳細

chromosome	start_position	end_position	Length(kb)	copy #	type	Gene	description
1	5730000	5740000		10	5	gain	none
1	40020000	40030000		10	0	loss	PABPC4
1	85980000	86010000		30	4	gain	DDAH1
1	144920000	144950000		30	5	gain	NBPF9
1	144990000	145090000		100	7	gain	PDE4DIP
1	145090000	145110000		20	6	gain	SEC22B
1	147710000	147730000		20	4	gain	NBPF8
1	156180000	156190000		10	6	gain	SLC25A44
1	229810000	229820000		10	1	loss	none
1	234910000	234920000		10	5	gain	none
1	248570000	248580000		10	5	gain	OR2T1
2	89830000	89890000		60	6	gain	none
2	133020000	133040000		20	16	gain	retro-GDC27
2	133050000	133060000		10	4	gain	none
3	4720000	4810000		90	1	loss	ITPR1
3	59820000	59910000		90	4	gain	FHIT
3	60180000	60370000		190	1	loss	FHIT
3	75760000	75790000		30	5	gain	ZNF717
3	196620000	196630000		10	5	gain	SENP5
4	50000	70000		20	9	gain	ZNF595
4	3580000	3640000		60	3	gain	LINC00955
4	10210000	10230000		20	0	loss	none
4	34780000	34830000		50	0	loss	none
4	68260000	68270000		10	15	gain	none
4	190470000	190680000		210	6	gain	none
5	21530000	21570000		40	4	gain	GUSBP1
5	53670000	127680000		74010	1	loss	many
5	128170000	135610000		7440	1	loss	many
5	135840000	139580000		3740	1	loss	many
5	156820000	156900000		80	1	loss	ADAM19
6	260000	370000		110	3	gain	DUSP22
6	31280000	31290000		10	0	loss	HLA-B
6	32450000	32660000		210	1	loss	HLA-DRB1
6	35750000	35770000		20	4	gain	CLPS
6	57210000	57370000		160	4	gain	PRIM2
6	57370000	57560000		190	6	gain	PRIM2
6	74590000	74600000		10	0	loss	none
6	126100000	126160000		60	1	loss	NCOA7
6	135280000	135810000		530	3	gain	MYB
6	157730000	157740000		10	4	gain	TMEM242
6	162720000	162910000		190	3	gain	PARK2
7	12840000	13510000		670	3	gain	retro-GDC27
7	57940000	58000000		60	3	gain	none
7	61540000	61560000		20	4	gain	none
7	61740000	61980000		240	5	gain	none
7	61980000	62040000		60	3	gain	none
7	130810000	131000000		190	1	loss	MKLN1
7	133780000	133800000		20	1	loss	none
7	152100000	152120000		20	7	gain	KMT2C
7	155120000	155180000		10	4	gain	none
8	2200000	2280000		80	4	gain	none
8	32680000	32690000		10	0	loss	none
8	43080000	43100000		10	4	gain	none
8	123280000	123310000		30	1	loss	none
8	126220000	126550000		330	16	gain	NSMCE2
8	126710000	127390000		680	16	gain	LINC00861
8	128060000	128070000		10	4	gain	none
8	128070000	128350000		280	16	gain	retro-SRRM1
8	128680000	128690000		10	4	gain	none
8	128690000	128770000		80	16	gain	MYC
8	128770000	128780000		10	5	gain	retro-RPL39
8	130000000	130090000		90	33	gain	none
8	130090000	130220000		130	17	gain	none
8	130360000	130700000		340	18	gain	CCDC26
9	11780000	32400000		20620	1	loss	many
9	66840000	66850000		10	7	gain	none
9	104630000	106950000		2320	1	loss	CYL2
9	107060000	107100000		40	1	loss	retro-WDR45B
9	130890000	130900000		10	4	gain	PTGES2
10	0	37870000		37870	1	loss	many
10	38780000	38820000		40	3	gain	none
10	38870000	39150000		280	3	gain	retro-PABPC1
10	42400000	42410000		10	63	gain	none
10	46970000	47150000		180	4	gain	ANXA8

Table 6 (つづき)

chromosome	start_position	end_position	Length(kb)	copy #	type	Gene	description	
11	0	190000		190	1	loss	AL137655	cDNA DKFpZp434E2016
11	200000	45180000	44980	1	loss	many		almost whole short arm
11	48340000	48390000	50	3	gain	OR4C45		olfactory receptor, family 4, subfamily C, member 45
11	48810000	48820000	10	4	gain	none		
11	50720000	50770000	50	3	gain	none		
11	51570000	51580000	10	24	gain	none		alpha satellite
11	54950000	54990000	40	1	loss	none		
11	55010000	55020000	10	3	gain	none		
11	85190000	85200000	10	4	gain	DLC2		discs, large homolog 2 (Drosophila)
12	2360000	2370000	10	4	gain	CACNA1C		calcium channel, voltage-dependent, L type, alpha 1C subunit
12	11220000	11240000	20	1	loss	PRR1-PRR4		PRR1-PRR4 readthrough
12	34830000	34840000	10	4	gain	none		alpha satellite
13	55460000	63620000	8160	3	gain	PCDH17他		protocadherin 17
13	63620000	63630000	10	6	gain	none		
13	63630000	63650000	20	5	gain	none		
13	63650000	115169878	51519.878	3	gain	many		half distal long arm
14	0	20430000	20430	3	gain	many		whole short arm +alpha
14	62180000	62630000	450	1	loss	HIF1A他		hypoxia inducible factor 1
14	64720000	81910000	17190	1	loss	many		middle of long arm
14	106540000	106560000	20	1	loss	abParts		immunoglobulin G heavy chain variable region
14	106710000	106730000	20	3	gain	abParts		immunoglobulin G heavy chain variable region
16	15050000	15060000	10	4	gain	none		
16	21570000	21590000	20	3	gain	LOC23117		cDNA FLJ59829 complete cd
16	22630000	22710000	80	3	gain	none		
16	32520000	32560000	40	5	gain	none		
16	32560000	33910000	1350	3	gain	TP53TG3他		TP53 target 3
16	33950000	33960000	10	21	gain	none		
16	34190000	34200000	10	4	gain	none		alpha satellite
16	34470000	34760000	290	3	gain	many		retroposed genes
16	35200000	35240000	40	3	gain	none		alpha satellite
16	70890000	71200000	310	4	gain	HYDIN		axonemal central pair apparatus protein
16	78270000	78320000	50	3	gain	WVVOX		VW domain containing oxidoreductase
16	81280000	81900000	620	1	loss	OMP1他		c-Maf inducing protein
16	85460000	90354753	4894.753	1	loss	many		
17	0	7490000	7490	1	loss	many		
17	7490000	7580000	90	0	loss	TP53		tumor protein p53
17	7580000	12820000	5240	1	loss	many		middle of short arm
17	12820000	12850000	30	0	loss	ARHGAP44		Rho GTPase activating protein 44
17	12850000	20680000	7830	1	loss	many		middle of short arm
17	21200000	21330000	130	4	gain	MAP2K3		mitogen-activated protein kinase kinase 3
17	34450000	34480000	30	3	gain	retro-GOLM1		golgi membrane protein 1
17	41390000	41400000	10	4	gain	none		
17	41400000	41410000	10	8	gain	none		
17	44210000	44280000	70	4	gain	KANSL1		KAT8 regulatory NSL complex subunit 1(histon acetylation)
17	44340000	44350000	10	6	gain	none		
17	45210000	45220000	10	4	gain	CCD27		cell division cycle 27
17	45220000	45250000	30	3	gain	CCD27		cell division cycle 27
18	2350000	2810000	460	3	gain	many		
18	9080000	9950000	870	3	gain	many		
18	45340000	45500000	160	3	gain	SMAD2		SMAD family member 2
18	46560000	46980000	420	3	gain	ZBTB7C		zinc finger and BTB domain containing 7C also CTIF & SMAD7
18	61430000	61800000	370	3	gain	SERPINF8		serpin peptidase inhibitor, clade B (ovalbumin
18	69200000	69310000	110	1	loss	LOC100505771		uncharacterized LOC100505776
19	20600000	20690000	90	1	loss	ZNF826P		zinc finger protein 826, pseudogene
19	24510000	24570000	60	4	gain	none		alpha satellite
19	24570000	27860000	3290	3	gain	none		centromere
19	27870000	27890000	20	7	gain	none		alpha satellite
19	27890000	27930000	40	3	gain	none		alpha satellite
20	1560000	1580000	20	0	loss	SIRPB1		signal-regulatory protein beta 1
20	14360000	14390000	30	1	loss	MACROD2		MACRO domain containing 2 (O-acetyl-ADP-ribose deacetylase)
20	14840000	15110000	270	1	loss	MACROD2		MACRO domain containing 2 (O-acetyl-ADP-ribose deacetylase)
20	16740000	16750000	10	1	loss	none		
20	26260000	26320000	60	3	gain	none		alpha satellite
20	29810000	29830000	20	4	gain	none		(GAATG)n
20	58420000	58500000	80	3	gain	SYCP2		synaptonemal complex protein 2
21	0	10710000	10710	7	gain	many		almost short arm
21	10710000	10760000	50	20	gain	none		L1PA3 repeat alpha satellite
21	10760000	10860000	100	7	gain	none		L1PA3 repeat alpha satellite
22	16850000	16860000	10	3	gain	none		HSATIII repeat
22	24380000	24390000	10	1	loss	GSTT1		glutathione S-transferase theta 1
22	25750000	25900000	150	3	gain	LRPSL		low density lipoprotein receptor-related protein 5-like

Table 7 17 番染色体上の遺伝子欠失領域



Invisible deletions were detected!

chromosome	start	end	size(kb)	copy #	type	Gene	description
17	0	7490000	7490	1	loss	many	
17	7490000	7580000	90	0	loss	TP53	tumor protein p53
17	7580000	12820000	5240	1	loss	many	middle of short arm
17	12820000	12850000	30	0	loss	ARHGAP44	Rho GTPase activating protein 44
17	12850000	20680000	7830	1	loss	many	middle of short arm
17	21200000	21330000	130	4	gain	MAP2K3	mitogen-activated protein kinase kinase 3
17	34450000	34480000	30	3	gain	retro-GOLM1	golgi membrane protein 1
17	41390000	41400000	10	4	gain	none	
17	41400000	41410000	10	8	gain	none	
17	44210000	44280000	70	4	gain	KANSL1	KAT8 regulatory NSL complex subunit 1
17	44340000	44350000	10	6	gain	none	
17	45210000	45220000	10	4	gain	CDC27	cell division cycle 27
17	45220000	45250000	30	3	gain	CDC27	cell division cycle 27

Figure 14 ホールゲノムシーケンスデータの冗長度

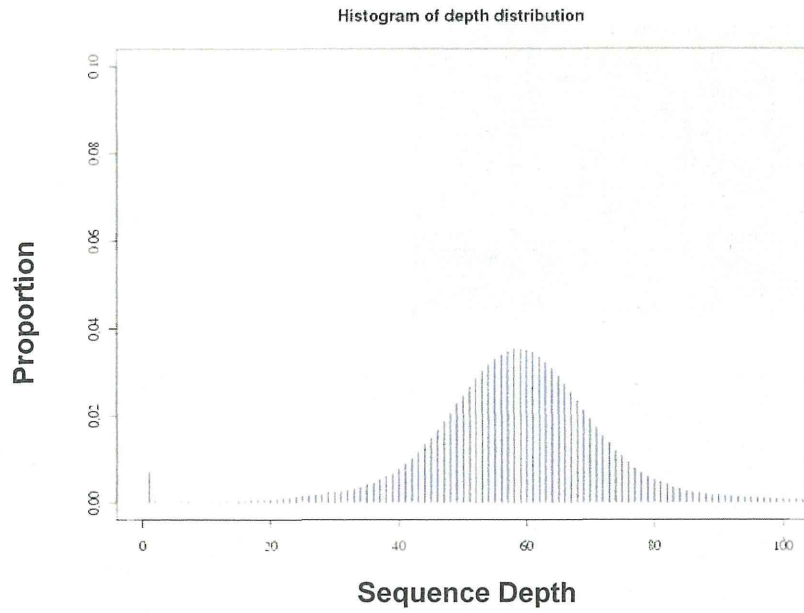


Figure 15 hMSC 細胞のシーケンスデータの冗長度を用いた CGH (コピー数異常) 解析

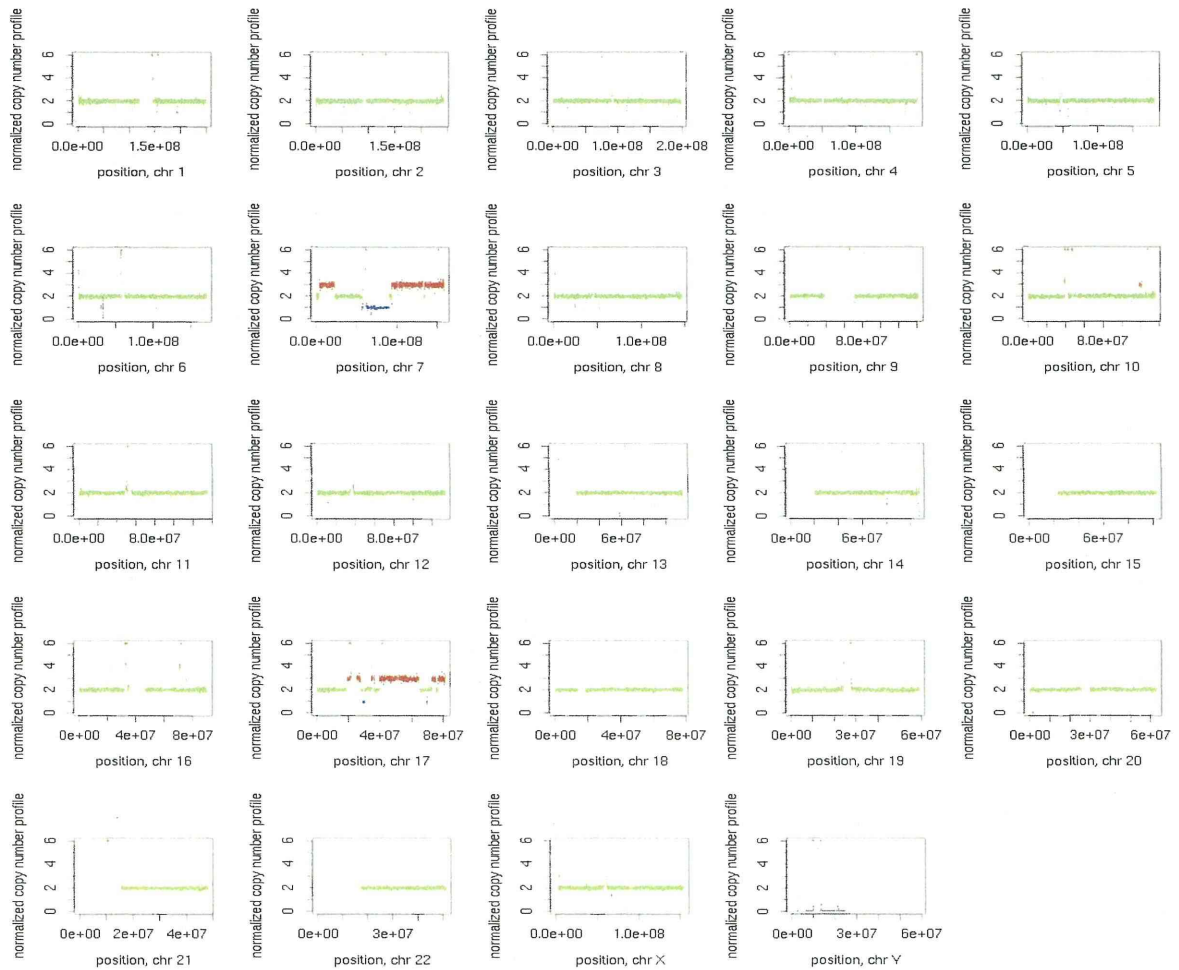
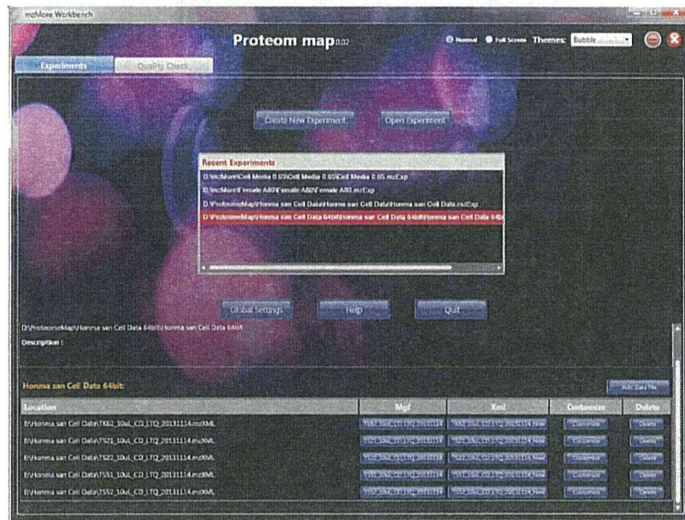


Figure 16 ProteomeMap ソフトウェアの概要

1) 生データの取り込み画面

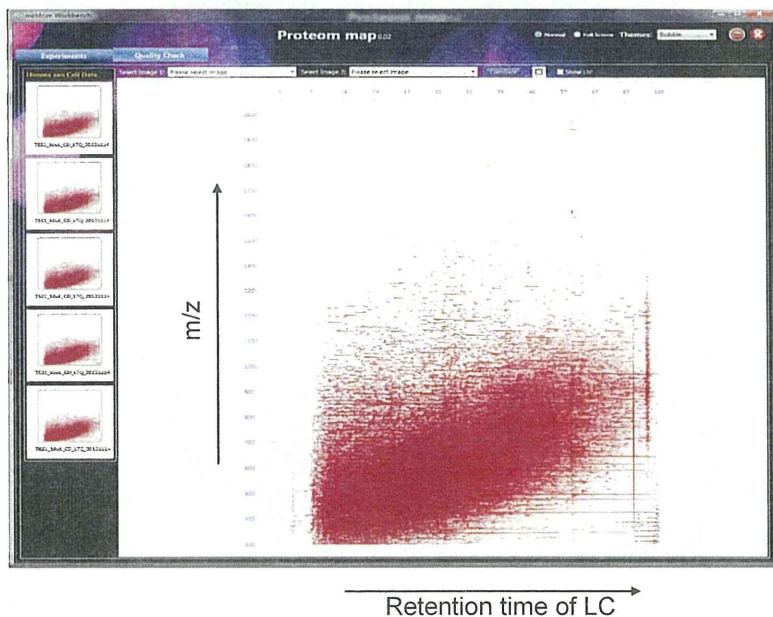
ProteomeMap

- put your proteins on the map...



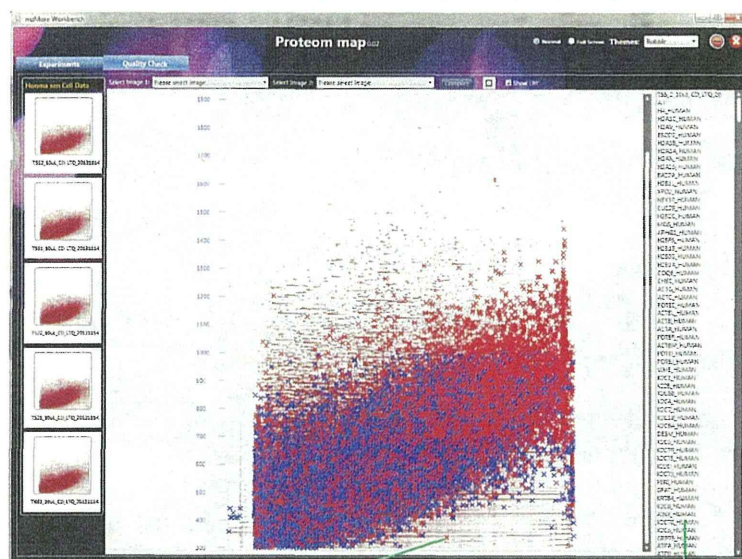
Compare
 what proteins are identified,
 where it is identified and
 how its identified
all in one screen

2) LC-MSデータの2次元マップ表示



3) MS/MSデータの取り込みと表示

Mapping of MS/MS Data on 3D Map & List of Identified Proteins



Location of MS/MS and identified proteins

List of identified proteins

4) ズームイン機能

Zoom IN Option of ProteomeMap



Blue X marks identified peptides

Red X marks un-identified MS/MS data

5) MS/MS スペクトルの表示

MS/MS View Option in ProteomeMap

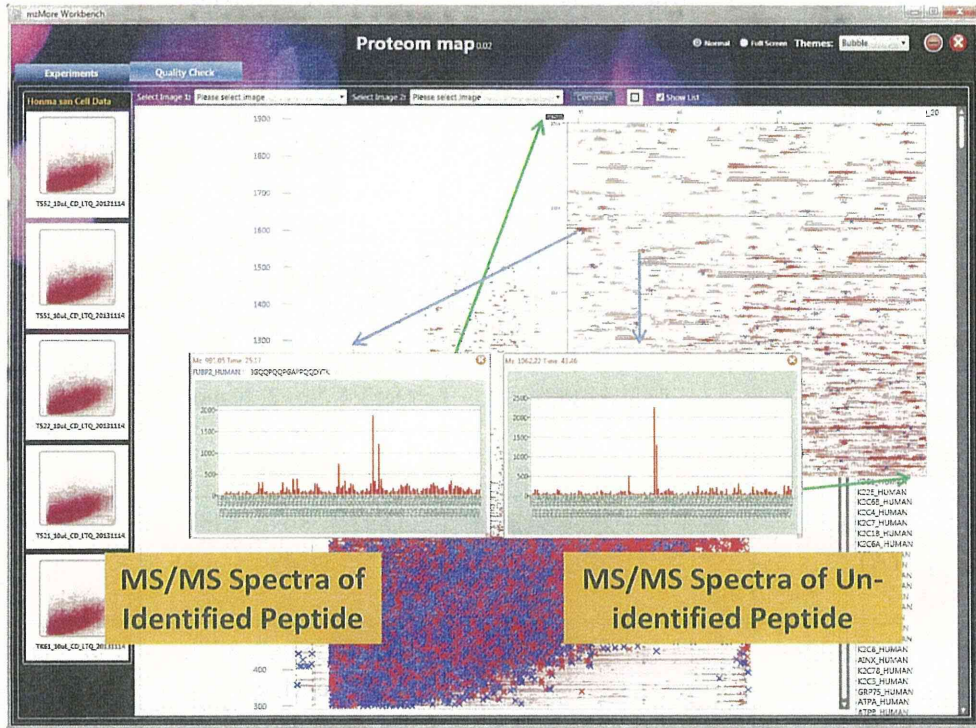


Table 8 本研究で用いたヒト iPS 細胞株

iPS 細胞株名	由来細胞	導入遺伝子	導入方法
201B7	皮膚線維芽細胞	Oct3/4, Sox2, Klf4, c-Myc	レトロウイルス
253G1	皮膚線維芽細胞	Oct3/4, Sox2, Klf4	レトロウイルス
409B2	皮膚線維芽細胞	Oct3/4, Sox2, Klf4, L-Myc, Lin28, p53shRNA	エピゾーマルベクター
Human mc-iPS (mc-iPS)	脂肪幹細胞	Oct3/4, Sox2, Nanog, Lin28	プラスミド
HiPS-RIKEN-1A (R-1A)	臍帯由来線維芽細胞	Oct3/4, Sox2, Klf4, c-Myc	レトロウイルス
HiPS-RIKEN-2A (R-2A)	臍帯由来線維芽細胞	Oct3/4, Sox2, Klf4, c-Myc	レトロウイルス
HiPS-RIKEN-12A (R-12A)	臍帯由来線維芽細胞	Oct3/4, Sox2, Klf4	レトロウイルス
Tic	肺線維芽細胞	Oct3/4, Sox2, Klf4, c-Myc	レトロウイルス
ATCC-DYR0110 hiPSc (Ai-100)	皮膚線維芽細胞	Oct3/4, Sox2, Klf4, c-Myc	レトロウイルス
ATCC-DYR0103 hiPSc (Ai-103)	肝線維芽細胞	Oct3/4, Sox2, Klf4, c-Myc	レトロウイルス

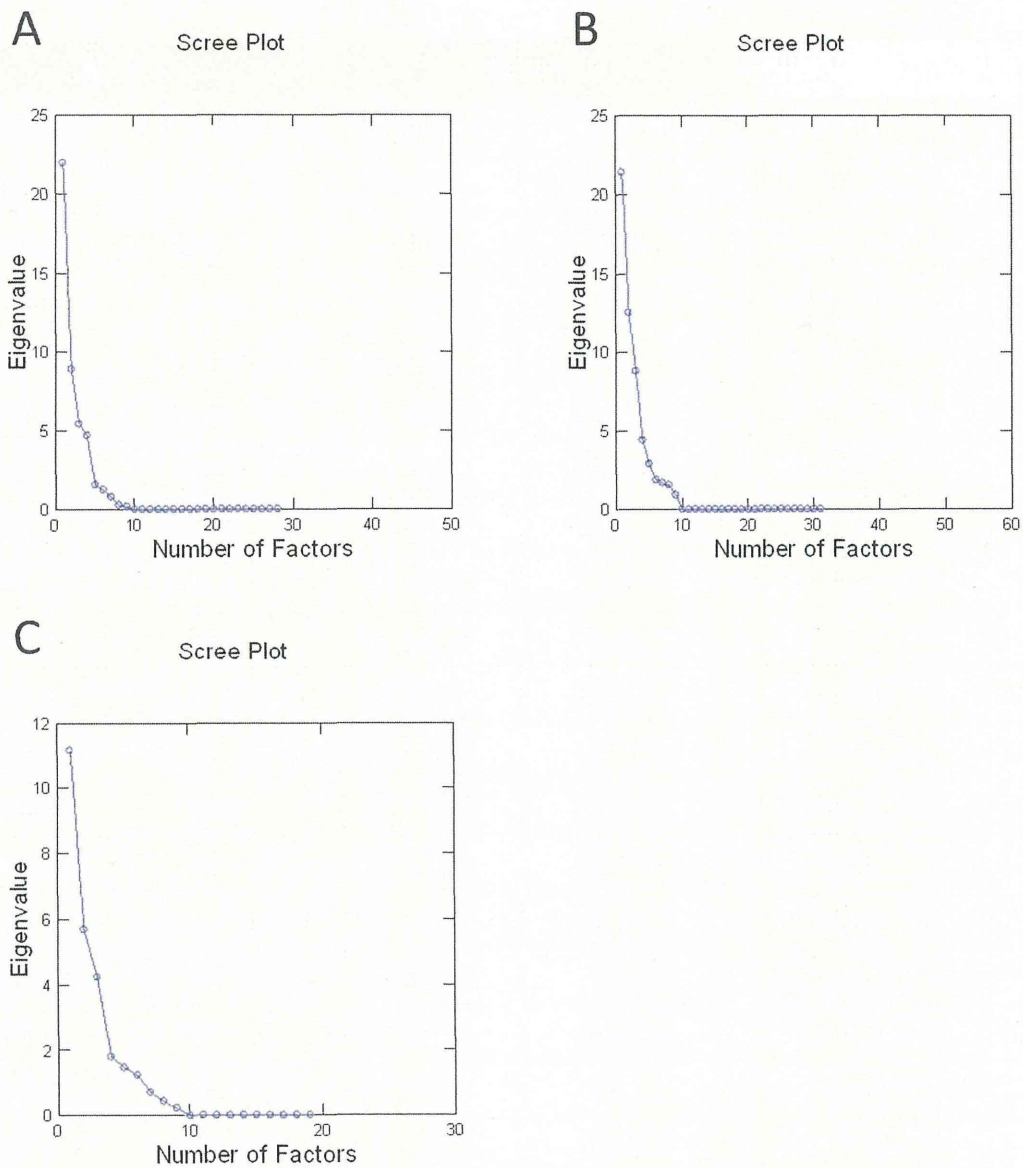


Figure 17 三胚葉系細胞マーカー遺伝子発現量による主成分分析

A) 外胚葉系細胞マーカーでの主成分寄与率, B) 中胚葉系細胞マーカーでの主成分寄与率, C) 内胚葉系細胞マーカーでの主成分寄与率.

Table 9 外胚葉, 中胚葉および内胚葉系細胞における遺伝子発現量の主成分分析から算出された第一主成分係数

外胚葉		中胚葉		内胚葉	
遺伝子名	主成分係数	遺伝子名	主成分係数	遺伝子名	主成分係数
<i>PAX3</i>	0.987	<i>TNFRSF1A</i>	0.969	<i>AFP</i>	0.948
<i>MAP2</i>	0.963	<i>ITGB3</i>	0.950	<i>GATA6</i>	0.931
<i>NES</i>	0.960	<i>ANPEP</i>	0.934	<i>GATA1</i>	0.907
<i>CDH2</i>	0.958	<i>STAT3</i>	0.928	<i>HNF1A</i>	0.904
<i>TUBB3</i>	0.955	<i>KDR</i>	0.926	<i>APOE</i>	0.894
<i>SYP</i>	0.954	<i>PDGFRA</i>	0.903	<i>CD44</i>	0.854
<i>PAX7</i>	0.949	<i>CD44</i>	0.889	<i>GATA4</i>	0.831
<i>PAX6</i>	0.915	<i>ITGB1</i>	0.888	<i>ITGB1</i>	0.795
<i>MAPT</i>	0.912	<i>MME</i>	0.882	<i>THY1</i>	0.794
<i>SOX9</i>	0.899	<i>PECAMI</i>	0.882	<i>ITGA6</i>	0.710
<i>SOX10</i>	0.884	<i>SPI1</i>	0.865	<i>GCG</i>	0.701
<i>NGFR</i>	0.854	<i>CD34</i>	0.855	<i>CDX2</i>	0.672
<i>OTX2</i>	0.854	<i>HAND1</i>	0.850	<i>SOX17</i>	0.598
<i>FOXD3</i>	0.851	<i>ITGAM</i>	0.844	<i>SLC2A2</i>	0.582
<i>NEFL</i>	0.836	<i>THY</i>	0.843	<i>SST</i>	0.535
<i>GBX2</i>	0.818	<i>ITGAL</i>	0.828	<i>SOX7</i>	0.533
<i>CRABP2</i>	0.808	<i>CD36</i>	0.823	<i>PDX1</i>	0.433
<i>MNX1</i>	0.798	<i>GATA4</i>	0.781	<i>NEUROG3</i>	0.398
<i>ITGA4</i>	0.787	<i>ITGA6</i>	0.781	<i>FOXA2</i>	0.394
<i>NCAM1</i>	0.785	<i>CEACAM1</i>	0.753	<i>CTNNB1</i>	0.364
<i>SOX2</i>	0.731	<i>MYOG</i>	0.738	<i>MIXL1</i>	0.201
<i>MCAM1</i>	0.650	<i>INHBA</i>	0.726	<i>HNF1B</i>	0.186
<i>NOTCH1</i>	0.645	<i>ITGAX</i>	0.647	<i>EOMES</i>	0.015
<i>TH</i>	0.516	<i>RUNX1</i>	0.620	<i>ISL1</i>	-0.399
<i>EN1</i>	0.455	<i>TWIST1</i>	0.614	<i>CDH2</i>	-0.438
<i>FAS</i>	0.391	<i>BMP2</i>	0.597	<i>SYP</i>	-0.463
<i>FGF5</i>	0.363	<i>ICAM1</i>	0.597	<i>PAX6</i>	-0.530

<i>NEUROG3</i>	0.361
<i>NOG</i>	0.239
<i>IRF6</i>	0.076
<i>FUT4</i>	-0.186
<i>PDGFRA</i>	-0.263
<i>ICAM1</i>	-0.271
<i>ITGB1</i>	-0.334
<i>FGFR2</i>	-0.378
<i>SNAI2</i>	-0.432
<i>ITGA6</i>	-0.454
<i>THY1</i>	-0.492
<i>GATA2</i>	-0.526
<i>HAND1</i>	-0.543
<i>GATA3</i>	-0.551
<i>ABCG2</i>	-0.597
<i>CD44</i>	-0.599
<i>APOE</i>	-0.672
<i>TDGF1</i>	-0.842

<i>ITGAV</i>	0.556
<i>ABCG2</i>	0.505
<i>SDC</i>	0.503
<i>FUT4</i>	0.457
<i>SRF</i>	0.455
<i>NODAL</i>	0.451
<i>CDH5</i>	0.429
<i>MYOD1</i>	0.419
<i>GATA2</i>	0.356
<i>KIT</i>	0.320
<i>CD4</i>	0.319
<i>GATA3</i>	0.306
<i>MIXL1</i>	0.271
<i>ADIPOQ</i>	0.270
<i>NOTCH1</i>	0.266
<i>T</i>	0.258
<i>ACTC1</i>	0.152
<i>EOMES</i>	0.077
<i>HHEX</i>	0.074
<i>NCAM1</i>	0.023
<i>FGF5</i>	0.008
<i>MCAM1</i>	-0.024
<i>ITGA4</i>	-0.070
<i>FOXC1</i>	-0.071
<i>NGFR</i>	-0.114
<i>LEF1</i>	-0.186
<i>NES</i>	-0.358
<i>CDH2</i>	-0.421
<i>DLL1</i>	-0.598