

A

Line-1 global

Line 1 elements (accession number X52235) Wild type sequence

61 **CG**cagaagac ggggtgatttc tgcatttoca tctgaggtac tgggttcac tcactagggg
121 gtgccagaca gtggg**CG**cag gtcagtggtt g**CGCG**cac**CG** tgca**CG**agc**C**Gaagcaggg**C**
181 **G**aggcattgt ctcaactggg aag**CG**caagg ggtcagggag ttccctttc**C**Gagttcaaga
241 aaggggtga**C**Gga**CG**cacct ggaaaat**CG**g gtctctcca cc**CG**aatatt g**CG**cttt**CG**
301 ac**CG**gcttaa aaaa**CG**g**CG**c ac**CGCG**agat tatactctgc acctggctag gagggtccta
361 **CG**ccca**CG**ga gtct**CG**ctga ttgtagcac agcagttctga gatcaaactg caagg**CG**ca
421 gcaaggctgg gggagggg**CG** cc**CG**ccattg ccaggcttg cttgggtaaa caaagcagcc
481 tggcagct**CG** aactgggtgg agcccaccac agctcaagga ggctgctg cttttgtagg

Line 1 elements (accession number X52235) Bisulfite sequence

61 **CG**tagaagat ggggtgatttt t**g**tattttta ttgaggtat tgggttt**tatt ttattagga** SN-9
121 **gtgtagata gtgg****CG**tag gttagtggtt g**CGCG**tat**CG** tgta**CG**agt**C**Gaagtaggg**C**
181 **G**agggtattgt tttattttggg aag**CG**taagg ggttagggag ttttttttt**C**Gagttaaaga
241 aaggggtga**C**Gga**CG**tattt ggaaaat**CG**g **gttttttta** tt**CG**aatatt g**CG**tttt**CG**
301 at**CG**gcttaa aaaa**CG**g**CG**t at**CGCG**agat tatactctgt atttggttag gagggtttta
361 **CG**ttta**CG**ga gttt**CG**ttga ttgtagtat agtagtttga gattaaattg taagg**CG**ta
421 gtaaggctgg gggagggg**CG** tt**CG**ttattg tttaggtttg tttgggtaaa taaagtagtt
481 tggtagtt**CG** a**attgggtgg agtttattat agtttaa**gga ggtttgtttg tttttgtagg
Bi-L1-F1
Bi-L1-R2

Pyrosequencing primer: SN-8; SIRPH Primer: SN-8 and SN-9

Supplementary Figure S1-A: LINE-1 global specific primers. CpG numbering is not continuous because of gaps in the consensus sequence when compared to the sequenced LINE-1 clones.

B

Alu Global

Alu (accession numbers: Alu-J U14567; Alu-Sb U14568; Alu-Sb1 U14569; Alu-Sb2 U14570; Alu-Sc U14571; Alu-Sp U14572; Alu-Sq U14573; Alu-Sx U14574)

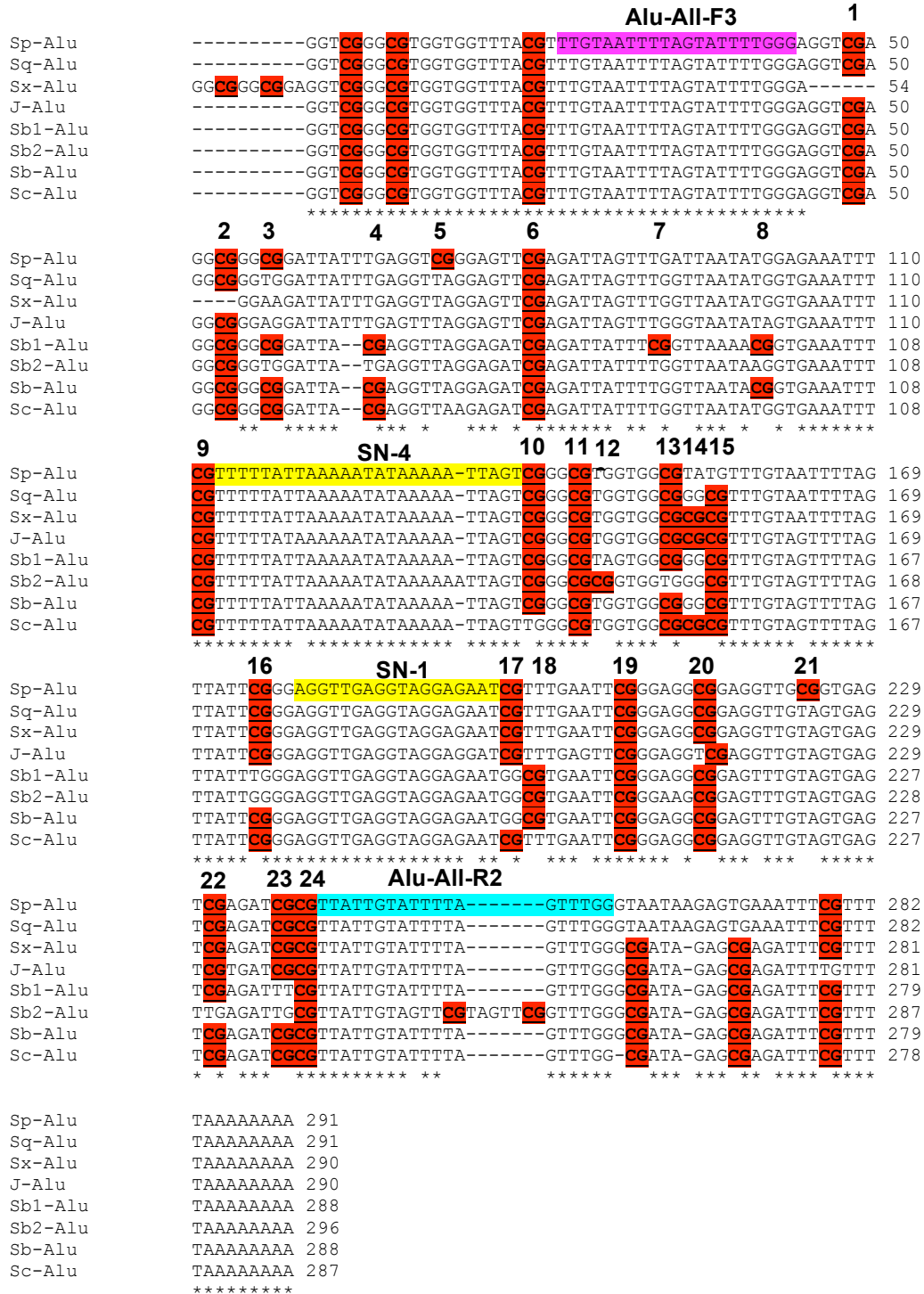
Wild type sequences alignment

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                                                                 1
Sp-Alu      -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
Sq-Alu      -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
Sx-Alu      GGCGGGCGGAGGCCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGA----- 54
J-Alu      -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
Sb1-Alu     -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
Sb2-Alu     -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
Sb-Alu      -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
Sc-Alu      -----GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGA 50
          *****
          2   3   4   5   6   7   8
Sp-Alu      GGCGGGCGGATCACCTGAGGTCGGGAGTTCGAGACCAGCCTGACCAACATGGAGAAACCC 110
Sq-Alu      GGCGGGTGGATCACCTGAGGTCGCAGGAGTTCGAGACCAGCCTGGCCAACATGGTGAACCC 110
Sx-Alu      ----GGAAGATCACCTGAGGTCGCAGGAGTTCGAGACCAGCCTGGCCAACATGGTGAACCC 110
J-Alu      GGCGGGAGGATCACTTGAGCCCGCAGGAGTTCGAGACCAGCCTGGCCAACATAGTGAACCC 110
Sb1-Alu     GGCGGGCGGATCA--CGAGGTCGCAGGAGATCGAGACCATCCCGGCTAAAACGGTGAACCC 108
Sb2-Alu     GGCGGGTGGATCA--TGAGGTCGCAGGAGATCGAGACCATCCTGGCTAACAAAGGTGAACCC 108
Sb-Alu      GGCGGGCGGATCA--CGAGGTCGCAGGAGATCGAGACCATCCTGGCTAACAACGGTGAACCC 108
Sc-Alu      GGCGGGCGGATCA--CGAGGTCAAGAGATCGAGACCATCCTGGCCAACATGGTGAACCC 108
          **  *****  ***  *  ***  *****  **  *  **  *****
          9   10  11  12  13  14  15
Sp-Alu      CGTCTCTACTAAAAATACAAAAA--TTAGCCGGGCGTGGTGGCGCATGCCTGTAATCCCAG 169
Sq-Alu      CGTCTCTACTAAAAATACAAAAA--TTAGCCGGGCGTGGTGGCGGGCGCCTGTAATCCCAG 169
Sx-Alu      CGTCTCTACTAAAAATACAAAAA--TTAGCCGGGCGTGGTGGCGGGCGCCTGTAATCCCAG 169
J-Alu      CGTCTCTACAAAAAATACAAAAA--TTAGCCGGGCGTGGTGGCGGGCGCCTGTAGTCCCAG 169
Sb1-Alu     CGTCTCTACTAAAAATACAAAAA--TTAGCCGGGCGTAGTGGCGGGCGCCTGTAGTCCCAG 167
Sb2-Alu     CGTCTCTACTAAAAATACAAAAAATTAGCCGGGCGGTGGCGGGCGCCTGTAGTCCCAG 168
Sb-Alu      CGTCTCTACTAAAAATACAAAAA--TTAGCCGGGCGTGGTGGCGGGCGCCTGTAGTCCCAG 167
Sc-Alu      CGTCTCTACTAAAAATACAAAAA--TTAGTGGGCGTGGTGGCGGGCGCCTGTAGTCCCAG 167
          *****  *****  *****  *****  *****  *****
          16  17  18  19  20  21
Sp-Alu      CTACGGGAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGAGGCGGAGGTTGCGGTGAG 229
Sq-Alu      CTACGGGAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 229
Sx-Alu      CTACGGGAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 229
J-Alu      CTACGGGAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 229
Sb1-Alu     CTACGGGAGGCTGAGGCAGGAGAATGGCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 227
Sb2-Alu     CTACGGGAGGCTGAGGCAGGAGAATGGCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 228
Sb-Alu      CTACGGGAGGCTGAGGCAGGAGAATGGCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 227
Sc-Alu      CTACGGGAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGAGGCGGAGGTTGCAGTGAG 227
          *****  *****  *****  *****  *****  *****
          22  23  24
Sp-Alu      CGGAGATCGCGCCACTGCACTCCA-----GCCTGGGCAACAAGAGCGAAACTCGCCTCT 282
Sq-Alu      CGGAGATCGCGCCACTGCACTCCA-----GCCTGGGCAACAAGAGCGAAACTCGCCTCT 282
Sx-Alu      CGGAGATCGCGCCACTGCACTCCA-----GCCTGGGCGACA-GAGCGGAGACTCGCCTCT 281
J-Alu      CGGTGATCGCGCCACTGCACTCCA-----GCCTGGGCGACA-GAGCGGAGACTCGCCTCT 281
Sb1-Alu     CGGAGATCCCGCCACTGCACTCCA-----GCCTGGGCGACA-GAGCGGAGACTCGCCTCT 279
Sb2-Alu     CGGAGATTGCGCCACTGCACTCCGCAGTCCCGCGCTGGGCGACA-GAGCGGAGACTCGCCTCT 287
Sb-Alu      CGGAGATCGCGCCACTGCACTCCA-----GCCTGGGCGACA-GAGCGGAGACTCGCCTCT 279
Sc-Alu      CGGAGATCGCGCCACTGCACTCCA-----GCCTGG-CGACA-GAGCGGAGACTCGCCTCT 278
          ***  ***  *****  *****  *****  *****
          *****  *****  *****  *****  *****  *****
Sp-Alu      CAAAAAAAAA 291
Sq-Alu      CAAAAAAAAA 291
Sx-Alu      CAAAAAAAAA 290
J-Alu      CAAAAAAAAA 290
Sb1-Alu     CAAAAAAAAA 288
Sb2-Alu     CAAAAAAAAA 296
Sb-Alu      CAAAAAAAAA 288
Sc-Alu      CAAAAAAAAA 287
          *****
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Supplementary Figure S1-B: Alu global specific primers (continued on next page)

Bisulfite sequences alignment



SIRPH primer: SN-1 and SN-4

Supplementary Figure S1-B: Alu global specific primers (continued from previous page)

C

Factor 8 region Xq28

Factor 8 (accession number: M88640) Wild type sequence

3061 agttcacatt tatcagaagg acctattccc taCGgaaact agcaatgggt ctcttgcca
3121 tctggatctCGGtgggaaggga gccttcttca gggaacagag ggagCGatta agtggaatga
3181 agcaaacaga cctggaaaag ttccctttct gagagtagca acagaaagct ctgcaaagac
3241 tccctccaag ctattggatc ctcttgcttg ggataaccac tatgggtactc agataccaaa
3301 agaagagtgg aatcccaag agaagtcacc agaaaaaaca gcttttaaga aaaaggatac
3361 cttttgtcc ctgaaCGctt gtgaaagcaa tcatgcaata gcagcaataa atgagggaca
3421 aaataagccCGGaaatagaag tcacctgggc aaagcaaggt aggactgaaa ggctgtgctc
3481 tcaaaacca ccagtcttga aaCGccatca aCGggaaata actCGtacta ctcttcagtc
3541 agatcaagag gaaattgact atgatgatac catatcagtt gaaatgaaga aggaagattt
3601 tgacatttat gatgaggatg aaaatcagag ccccCGcagc tttcaaaaga aaacaCGaca
3661 ctattttatt gctgcagtgg agaggctctg ggattatggg atgagtagct ccccatgt

Factor 8 (accession number: M88640) Bisulfite treated sequence

3061 agtttatatt tattagaagg atttatTTTT taCGgaaatt agtaatgggt tttttgggta
3121 tttggatttCGGtgggaaggga gtttttttta gggaaatagag ggagCGatta agtggaatga
3181 agtaaataga tttggaaaag tttttttttt gagagtagta atagaaagtt ttgtaaagat
3241 tttttttaag ttattggatt tttttgtttg ggataattat tatgggtattt agatattaaa
3301 agaagagtgg aaattttaag agaagttatt agaaaaaata gtttttaaga aaaaggatat
3361 tttttgttt ttgaaCGttt gtgaaagtaa ttatgtaata gtagtaataa atgagggata
3421 aaataagttCGGaaatagaag ttatttgggt aaagtaaggt aggattgaaa ggttgtgttt
3481 ttaaaattta ttagttttga aaCGttatta aCGggaaata attCGtatta ttttttagtt
3541 agattaagag gaaattgatt atgatgatat tatattagtt gaaatgaaga aggaagattt
3601 tgatatttat gatgaggatg aaaattagag ttttCGtagt ttttaaaaga aaataCGata
3661 ttattttatt gttgtagtgg agaggtttttgcattatggg atgagtagtt ttttatatgt
BiF8-F6 1 BiF8-F5
SN-1 7 8
BiF8-R5 BiF8-R6

Pyrosequencing primer: SN-1

Supplementary Figure S1-C: F8 region specific primers