

Additional File 2

Figure A. Fragment analysis of breakpoint PCR of the familial 1325-bp deletion encompassing exon 8 of the *STK11* gene.

Figure shows an agarose gel with the deleted allele preferentially amplified in the proband. Size ladder (Invitrogen 1 KB plus) is in the lane labeled as 1 kb+, while wild-type DNA in lane 1, proband DNA in lane 2, and water in lane 3. The two alternative forward primers used in amplification are labeled as Fa and Fb. The expected sizes of fragments generated from normal wild type alleles are written on top of the gel and the sizes of the amplicons generated from deleted alleles are highlighted with an arrow on the right. Asterisks highlight the bands sequenced.

Figure B. Sequence from the deletion locus of the the familial 1325-bp deletion encompassing exon 8 of the *STK11* gene.

Sequence shows the breakpoints in relation to the adjoining sequence; repeat elements, SNPs, exons, and primers. Sequence coordinates are listed according to UCSC hg 18 build (March 2006), and the two interruptions demarcate the breakpoints. Nucleotides in bold and capitalized font represent exons. Nucleotides in bold and underlined font correspond to the primers used in breakpoint PCR. SNPs (130 build) are highlighted in red font, and microhomology at breakpoints is shown boxed.

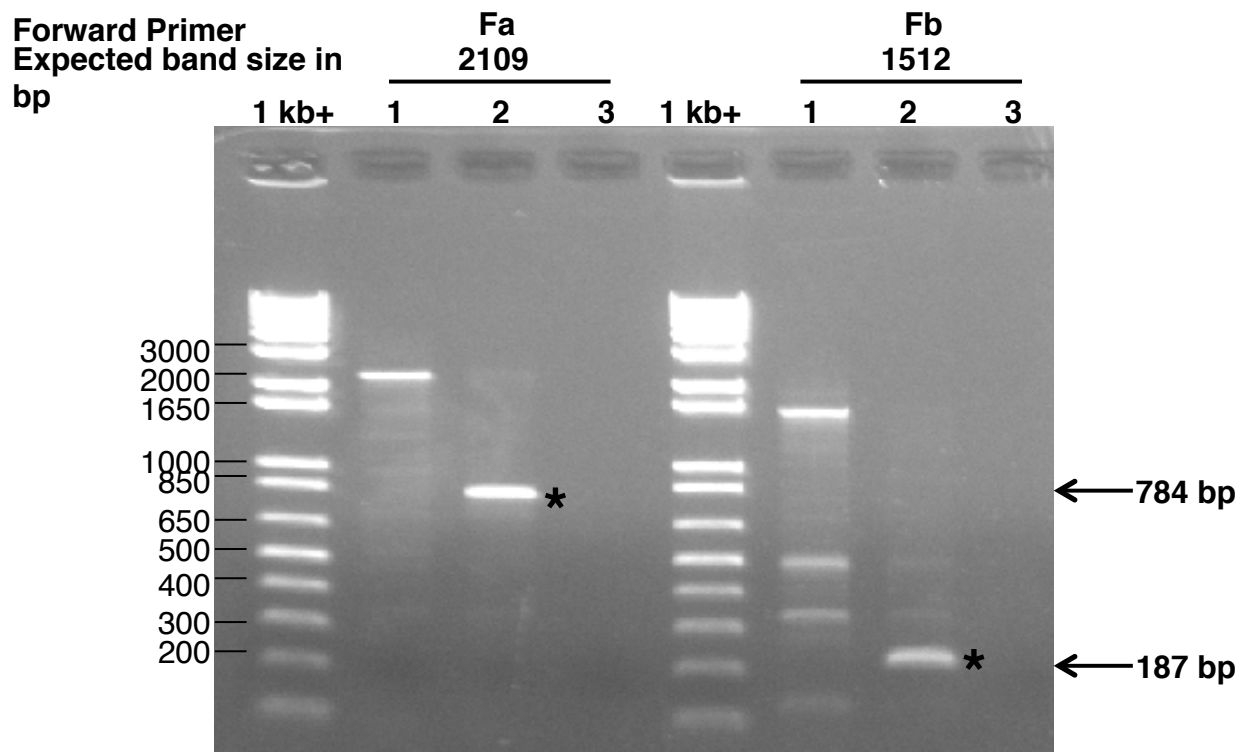
Figure C. Fragment analysis of breakpoint PCR of the familial 971-bp deletion encompassing exon 3 of the *STK11* gene.

Figure shows an agarose gel with the deleted allele preferentially amplified in the proband with one of four reverse primers used. Size ladder (Invitrogen 1 KB plus) is in the lane labeled as 1 kb+, while wild-type DNA in lane 1, proband DNA in lane 2, and water in lane 3. The four alternative reverse primers used in amplification are labeled as Ra, Rb, Rc and Rd. The expected sizes of fragments generated from normal wild type alleles are written on top of the gel. The size of the amplicon generated from deleted allele, when amplified using Rd primer, is highlighted with an arrow on the right. Asterisks highlight the bands sequenced.

Figure D. Sequence from the deletion locus of the the familial 97-bp deletion encompassing exon 3 of the *STK11* gene.

Sequence shows the breakpoints in relation to the adjoining sequence; repeat elements, SNPs, exons, and primers. Sequence coordinates are listed according to UCSC hg 18 build (March 2006), and the two interruptions demarcate the breakpoints. Nucleotides in bold and capitalized font represent exons. Nucleotides in bold and underlined font correspond to the primers used in breakpoint PCR. RepeatMasker are highlighted with blue font, SNPs (130 build) are highlighted in red font, and microhomology at breakpoints is shown boxed.

Additional File 2, Figure A



Additional File 2, Figure B

chr19:1173251-1174850

aggcatg**gagatgcgccaggaagg**gcacagctggtcccaaactggcga **Fb**
gagcctctcttttttc**c**cctcctcctggggctcccagcagcaggggtgtggc
tgggatccagcccagggcccccagctccatgacaggggaagacagagcagc
Ggacgggggtcagcag

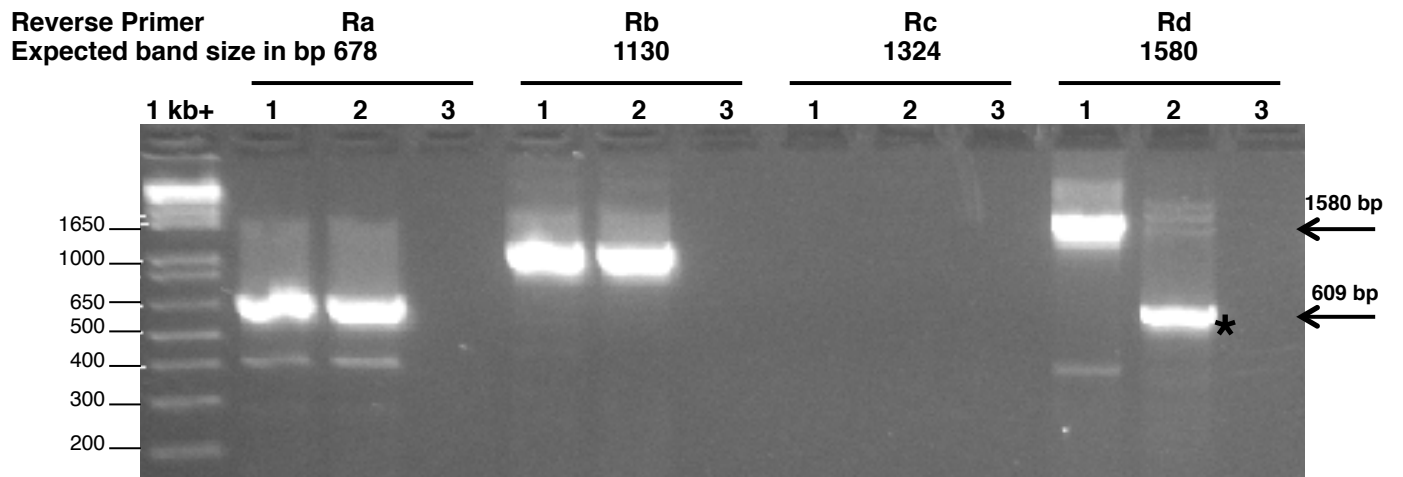
(Breakpoint @ chr19:1,173,416 in intron 7)

gccccacagtgcgcgctcctcacttc**g**tgggctc
tgctcctctgcaccagcccctggaggcccttgagccgtctgctggagccc
ctccgagccccgaggccaccactgagaccggctct**g**ggag**t**gggagtgt
ccggaccctgaggcgtggtgctgattgtgccttgggggtctctgcaca
gctcgggtcat**c**tgggcgctggcggggactggggctgcccccgatagc
ctcctgggctgggatgtgctcagggcccccagacccccttctggcctt
gctggctttgcagccagcatccatctggtgggtgctggcttctgagtgc
acctgggacacaggcctcaggggtggagg**g**gacatctgtcaggcttggagt
caggtcagcctgcctgctcctagaggacatggctgagcttctgtggtcac
agccacccttgacggcctggtcc**c**agctcctgagtgtgtggcaggtac
cctgggcccagaggagctgggtcggaaa**a**ctggaccgcccctggtgccagc
ctgacaggcgccactgcttctgggcgtttgag**CTGGTTCCGGAAGAAAC**
ATCCTCCGGCTGAAGCACCAGTGCCCATCCCACCGAGCCAGACACCAAG Exon 8
GACCGGTGGCGCAGCATGACTGTGGTGCCGTACTTGGAGGACCTGCACGG
CGCGGACGAGGACGAGGACCTCTTC**GACATCGAGGATGACATCATCTACA**
CTCAGGACTTCACGGTGCCCGgtgagtctggcgggggcccctgcccggct
ctgctgactcggccaggatgtcccacgggagcaggggtgctgctgctg
caaca-----390 bp not shown-----aggcg
tctgaggcagggccttagagcggagcgcggccttgacagaagagcagggctc
Agacctttcaggagaggaagcctctcggccggcg**c**gag

(Breakpoint @ chr19:1,174,740 in intron 8)

tagtgctga
ggaggagctcagggccttagcgtaggggcggcc**cacattggcagccagcc** **R**
cctccccgcatgctcccggcttggctgtgttcggcccagggctgggccc

Additional File 2, Figure C



Additional File 2, Figure D

chr19:1,169,801-1,171,400

cgggctgaccgttgaggccattttggtcgtaggctggggcgtgtcctcgtg **F**
tcacatctgtggacacccccatgggtcttacgggacagcctccctacgggg
actttgcttccctaaggccctgtgcccagagcaagagccagaagtggctcct
gaggctggggctgtgttccctgagccacgcggtcaggggcccctgggaccg
tcctgcatggggcccagcctgcttggggggcgtccaggaggcaccatcc
ccgcccattgggcagggtggg

(Breakpoint @ chr19:1,170,072 in intron 2)

ggacgtgagccccgcaggaacgctgcccc
aagagtcagccctgtcctccccctccccgtaggctccttctcctcctgggac
gctggggcccctgggccttttcagaggggtggctgagggcagggtgggccc
ctgggtcccagaggaggggcaaggtgggtgcagagggctccctccagagcccc
ttttctggccccctgtcctcctgggcctgtgagtggggccgccccctgag
ctgtgtgtccttagcgcccccacGTATATGGTGTGATGGAGTACTGCGTGTGT
GGCATGCAGGAAATGCTGGACAGCGTGCCGGAGAAGCGTTTCCCAGTGTG Exon 3

CCAGGCCACGGgtgctgtgcgcggggcagggggccagggtggggcgggggc
cgggggccaggcagggcaggctcctttccgtgaggccacactgctgtcc **Ra**

tgatattcattgacatgaaggcccaagttttttgtttttttgttttttt
gtgttttttttcgagatggagtctcactctgtcgcccaggctggagtgca
atggtgcatctcggctcactgcaagctccgcctccgaggttcacgccat **AluY**
tcttctgcctcagcttcccagtagctgggattacaggcgcccgccacca
cgcccggctaattttttgtatttttagtagagacggggtttcaccgtgtt
agccaggatgggtctcaaacctcctgacctcgtgatccgcctgcctcagcct
cccaaagtgtgggattacaggcatgagctaccacgcccggccttgtaaa

ggcccaagtttttaaaaacagttttgggggtcccccatgtgtggcatccac
aggcagggtgctgccaacctccgcctccatctttgctgaggcctgctgc **Rb**
ctgaggccagtggtcctgcttccagcccacgctggcagccgctgcctg
accagatctcctggatgcaggctgtgtggcctcagagtcaggg

(Breakpoint @ chr19:1171042 in intron 3)

cccttgcc
tgctgcaggaccacaggggcagggaggggcctgctgttccagcaagactt
tgggggtgcagccggcctgtgagccacaggaatgagacctgtggacatc **Rc**
cggggcccctgcccagacgtggctcggcgggacgaggggtggccactgcaggc
gcagggtgtggctcctgctggacctagccttctcctctgtcctgtgtgct
ggacttctgtgacttcccagctgggcctgtgggtgtttgggaggctcccag
gcagctgcaaaggggacccccgtgtagggggcagggaggcctcggcccagg
acgggtgtgtgctgcccgcagGTACTTCTGTCAGCTGATTGACGGCCTGG **Rd**