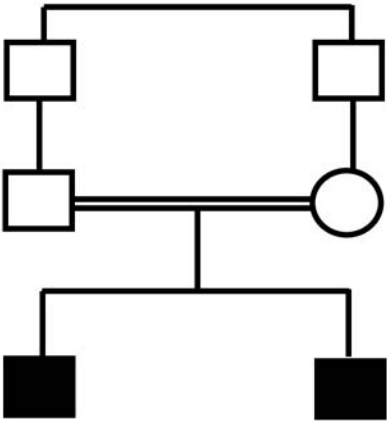


Figure S1. Pedigrees of the families affected with SMED-SL from UAE with the mutation found in the affected children of each family. In both families consanguinity (first cousin marriages) is evidenced.

A) Family1 [p.E113K]



B) Family 2 [p.R752C]

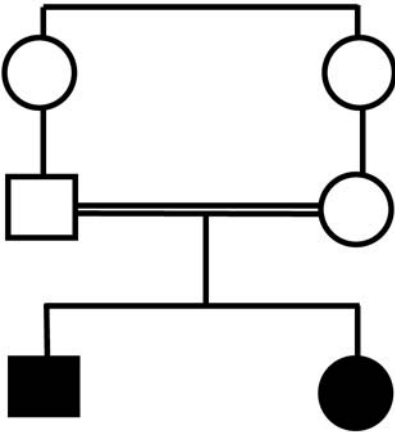
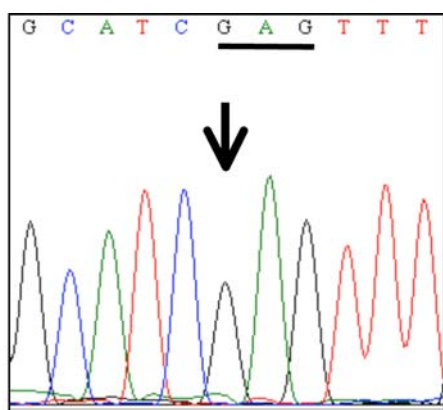
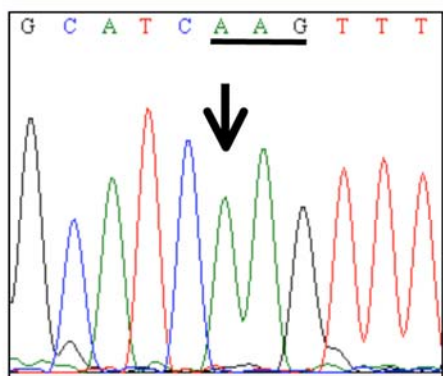


Figure S2. Chromatograms of DNA sequences showing the p.E113K (A) and p.R752C (B) mutations in patients with SMED-SL

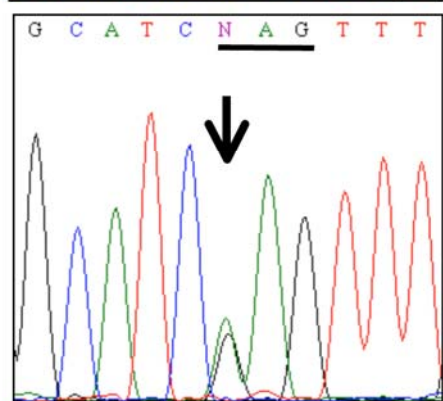
A) c.337G >A [p.E113K]



Control

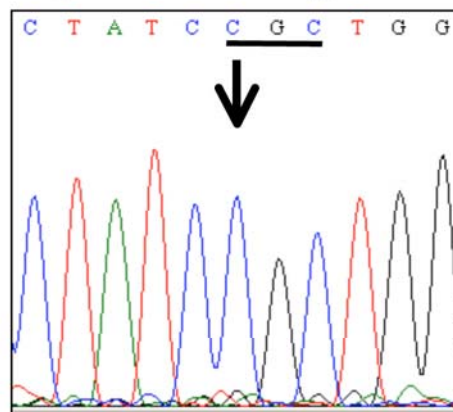


Patient

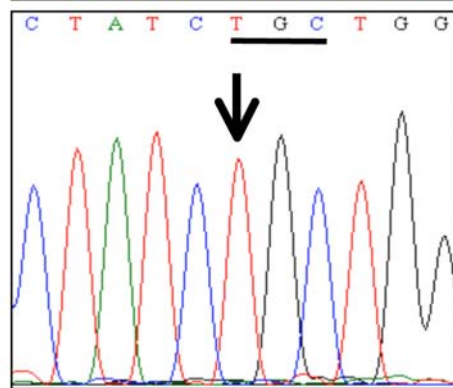


Parent

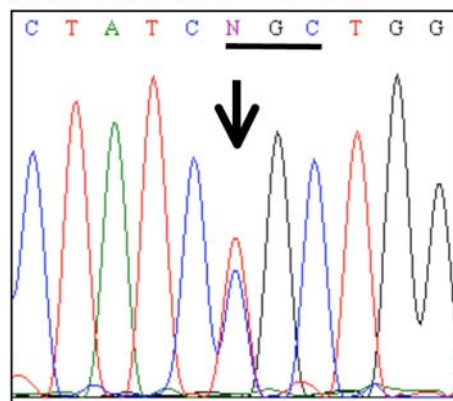
B) c.2254C >T [p.R752C]



Control



Patient



Parent

Figure S3. Alignment of DDR2 showing conservation of the E113 residue (boxed) in DDR2 proteins from different species

HumDDR2	HFITLVGTQGRHAGGHGIEFA	PMYKINYSRDG	TRWISWRNR
PanDDR2	HFITLVGTQGRHAGGHGIEFA	PMYKINYSRDG	TRWISWRNR
CowDDR2	HFITLVGTQGRHAGGHGIEFA	PMYKINYSRDG	TRWISWRNR
RatDDR2	HFITLVGTQGRHAGGHGIEFA	PMYKINYSRDG	NRWISWRNR
MusDDR2	HFITLVGTQGRHAGGHGIEFA	PMYKINYSRDG	SRWISWRNR
CanDDR2	HFITLVGTQGRHAGGHGIEFA	PMYKINYSRDG	TRWISWRNR
ZebDDR2	HFITLVGTQGRHAGGIGN	EFAQTYKIKYSRDG	SRWISWRNR

Table S1. The sequences of the primers used to amplify the coding exons and splice sites of the *DDR2* gene from genomic DNA.

Exon	Forward	Reverse
3	TGAGAATTGTACTIONCATTTCATGTTGG	GTAGTCCCTCTTGGCAGCTT
4	TCTTATTCCTTGTTCAATATTCAGTG	CCCCTAGGGTCAGGAATCTG
5	CAGCTGCTTGCCTGTGAAC	CACACAGAAAACCTGTACCCTTC
6	GTGGTGGGGTGAAGAAAAGT	TCCCTTTCTGATTTGATTGC
7	CGCTGTGCAAGCTTATACCC	TTGATTGATTATTGATCCCAAGA
8	GAGTGAAGATGCCGGGTAAA	TGAACTGGCATCAGCCTAGA
9	TACTGAGTTGGCTGGCACTG	TGAGAAGTTCTGGGCATGTG
10	TCACTAAATTGATCTTGTAATGTGC	CCAGGGCTACTCTTCATCCA
11	AGGAACAGGGTCTACCTCCA	AAATGTTTGCAATTTGCCTTTT
12	TGGGAGAGCTGAGTTTAAGAAGA	GCAGAGACTAAAAATAGATGCAATGA
13	GCCCTCCTCTCAGAGTTCCT	GTGAATCCACCTCTGGAAGG
14	GGAAATGCCAGCAAGAGTA	GCTCACTGACCTTCCCATCT
15	ATAGGCCTTGGTGTGCATTC	ACTGACTTCCCCCACCATC
16	GAATGTTGAGCTTTCAACCCTA	AGCCCACAAGCCAGTTGTTA
17	AGAATTCCTTGCCTGTGGTG	AGTGACAAAGACTAACACCTGGA
18	CAAATCAAACCATGATGCAAA	TGTCCAGATGGAGTGGCATA