SUPPORTING INFORMATION

Supp. Methods

Mutation analysis for *SMC1A* gene

DNA was extracted from peripheral blood lymphocytes by a standard non-organic extraction procedure. The entire *SMC1A* coding region was analysed for mutations in all patients by sequencing. Primer pairs were designed to amplify exons, exon/intron boundaries and short flanking intronic sequences. Amplified PCR products were purified (Qiagen) and sequenced. None of *SMC1A* mutations has been found in at least 100 control alleles.

Supp. Table S1. Polymorphisms in the non coding regions of *SMC1A* gene

Region	Nucleotide change	Reference
5' UTR	c.-19C>T	Deardorff et al. [2007]
Intron 1	c.109+101delG	NCBI
Intron 1	c.109+102A>C	NCBI
Intron 1	c.109+2743G>A	NCBI
Intron 1	c.109+3295T>C	NCBI
Intron 1	c.110-2132G>A	NCBI
Intron 1	c.110-352G>A	NCBI
Intron 1	c.110-169G>A	NCBI
Intron 3	c.411+71C>G	NCBI
Intron 7	c.1254+52A>G	NCBI
Intron 7	c.1254+973T>G	NCBI
Intron 7	c.1255-370C>T	NCBI
Intron 8	c.1338-59C>T	NCBI
Intron 8	c.1338-32C>A	Deardorff et al. [2007]
Intron 9	c.1545+792A>T	NCBI
Intron 9	c.1545+1356A>T	NCBI
Intron 9	c.1545+1379C>T	NCBI
Intron 9	c.1545+1416G>T	NCBI
Intron 9	c.1546-1002T>C	NCBI
Intron 9	c.1546-489G>T	NCBI
Intron 12	c.2058+22T>G	NCBI
Intron 12	c.2058+28C>T	NCBI
Intron 12	c.2058+29A>T	NCBI
Intron 13	c.2197-11A>T	This report
Intron 13	c.2197-5T>C	NCBI
Intron 15	c.2420+816G>A	NCBI
Intron 15	c.2420+828G>A	NCBI

Region	Nucleotide change	Reference
Intron 16	c.2562+1471C>T	NCBI
Intron 16	c.2562+1477T>G	NCBI
Intron 16	c.2562+1483T>C	NCBI
Intron 16	c.2562+2010delT	NCBI
Intron 18	c.2862+228C>T	NCBI
Intron 18	c.2862+387C>A	NCBI
Intron 18	c.2862+597C>T	NCBI
Intron 19	c.2973+1188G>A	NCBI
Intron 19	c.2973+3511C>T	NCBI
Intron 19	c.2973+3642C>T	NCBI
Intron 19	c.2973+4150G>A	NCBI
Intron 19	c.2973+4515T>G	NCBI
Intron 19	c.2973+5048delT	NCBI
Intron 19	c.2973+5816G>C	NCBI
Intron 19	c.2973+5950G>A	NCBI
Intron 19	c.2973+6655A>G	NCBI
Intron 19	c.2973+7619G>T	NCBI
Intron 19	c.2974-2169T>G	NCBI
Intron 19	c.2974-1096G>C	NCBI
Intron 20	c.3130+183G>A	NCBI
Intron 22	c.3438-94delA	This report
3’UTR	c.3702+315G>T	NCBI
3’UTR	c.3702+2443A>G	NCBI
3’UTR	c.3702+5016T>C	NCBI

Numbering of mutations based on GenBank NM_006306.2 with +1 as the A of the ATG start codon.