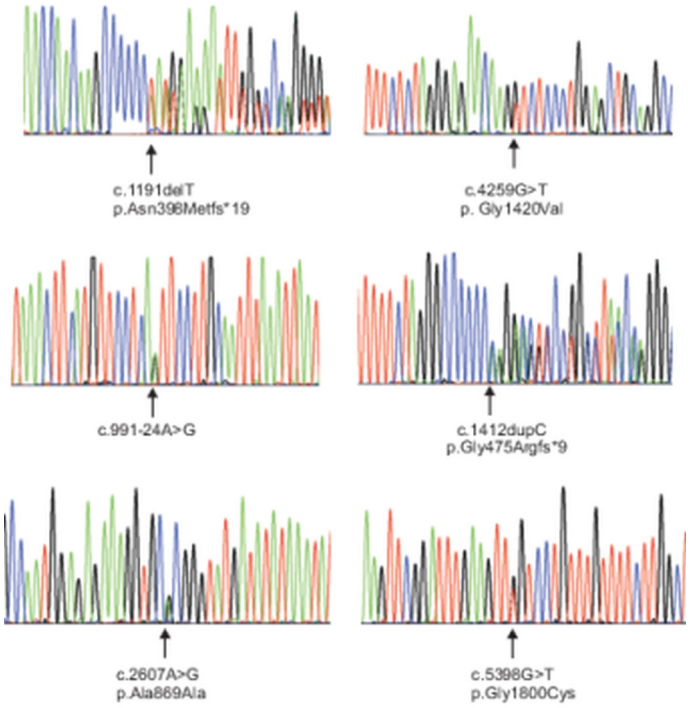
Supplemetary Figure 1. COL11A1 mutation sequences.

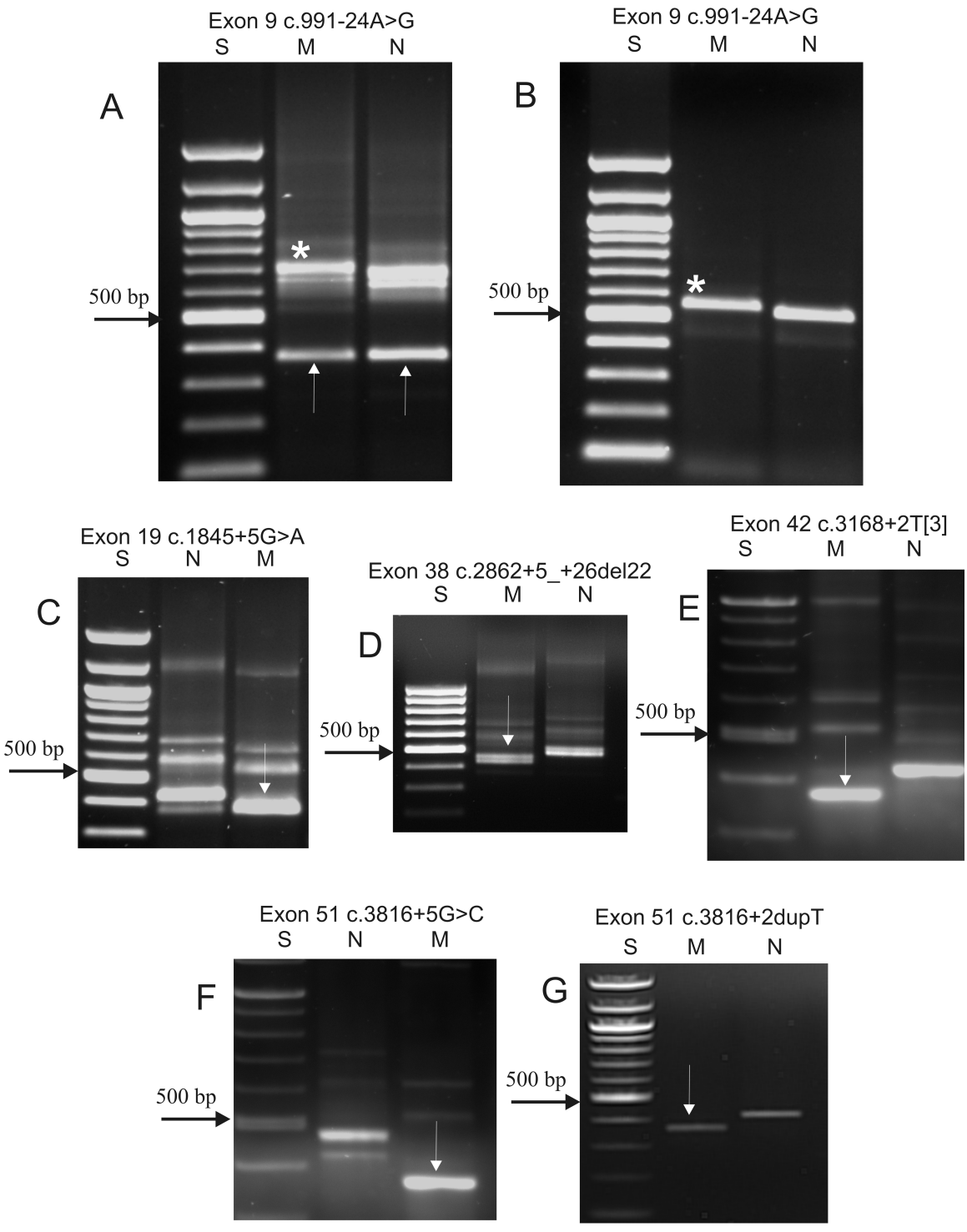
Variants found when sequencing individuals with Stickler syndrome. Green =A, Red=T, Blue=C, Black=G. The cDNA numbering uses the reference sequence NM_001854.3. where c.1 refers to the A of the initiating ATG codon for methionine which is numbered as p.1Met.



Supplementary Figure 1

Supplemetary Figure 2. Minigene analysis of *COL11A1* mutations.

Both mutant (M) and normal (N) *COL11A1* exon 9 alleles from an individual with the c.991-24A>G mutation were cloned into the splicing reporter USR13, transfected into MIO-M1 cells and analysed by RT-PCR and agarose gel electrophoresis using either vector specific primers (A) or a vector specific primer and a *COL11A1* exon 9 specific primer (B). Other mutations C-G (as indicated) from individuals with dominant type 2 Stickler syndrome were analysed in the same splicing reporter. Products representing exon skipped transcripts are indicated with arrows. An asterisk (*) indicates the product from the c.991-24A>G construct which is larger than the normal control. 100 bp size standards (S) are included for each reaction and the 500 bp standard indicated.



Supplementary Figure 2