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Supplemental Data

Mutations in *FKBP14* Cause a Variant of Ehlers-Danlos Syndrome with Progressive Kyphoscoliosis, Myopathy, and Hearing Loss

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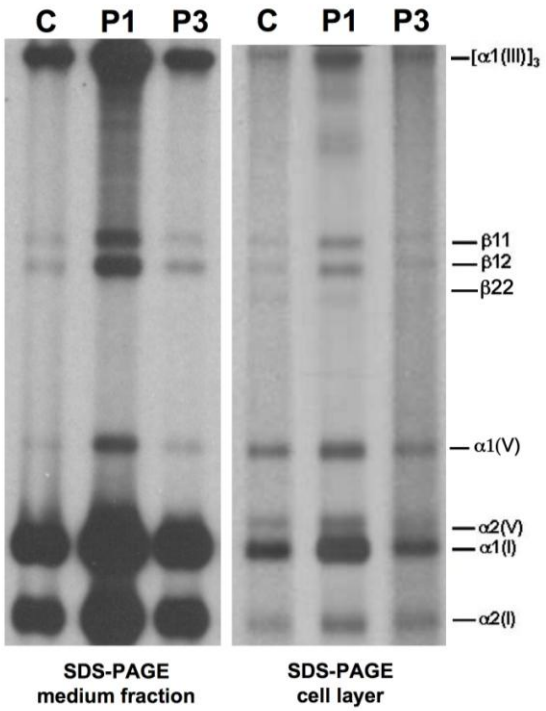

Figure S1. Collagen biochemical analysis
Pepsin-treated procollagens from medium and cell layers produced by fibroblasts from individuals P1 and P3 and a control (C) were electrophoresed on a 5% SDS-polyacrylamide gel, processed for fluorography, and exposed to X-ray films. The α1(I), α2(I), α1(V), α2(V), and $[\alpha1(III)]_3$ chains, and β components of type I collagens are indicated. Collagens of P1 and P3 migrate similarly to those from the control, and different from those of individuals with EDS VIA.

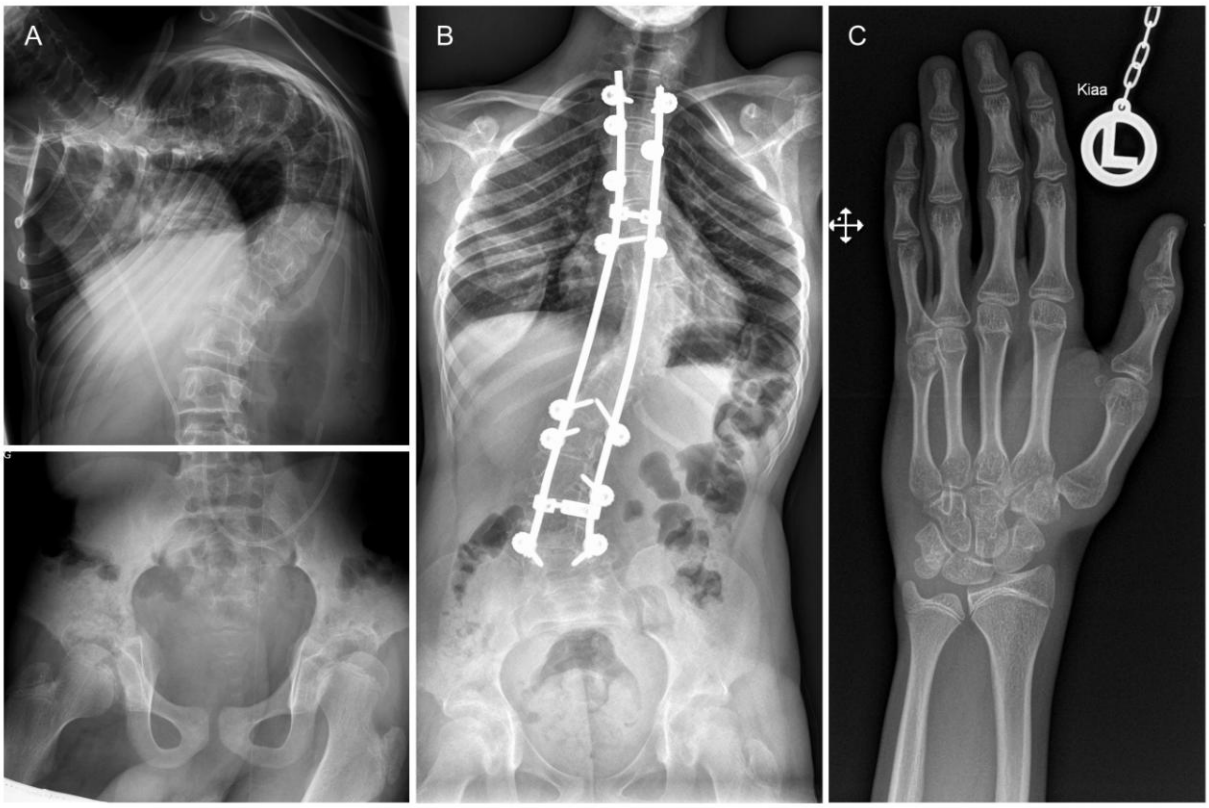

Figure S2. Radiographs

(A) Radiographs of the spine and the pelvis of P1 at the age of 13 years. Note the severe kyphoscoliosis and moderate osteopenia evidenced by translucent vertebral bodies in comparison to the cortical outline, deformed vertebral bodies, and trabecular prominence in femoral necks.
(B) Radiograph of the spine and the pelvis of P3 at the age of 11 years after surgical correction for kyphoscoliosis. Note the mild osteopenia evidenced by the accentuation of the cortical outline of the vertebral bodies.
(C) The radiograph of the left hand of P1 at the age of 16 years shows a slender shape due to the paucity of muscles and fat; the metaphyses and epiphyses are somewhat broad and flat, respectively. Bone age corresponds to chronological age.

Table S1. Primer sequences for sequence analysis of *FKBP14*

primer name	sequence (5‘- 3‘)	PCR fragment length
FKBP14 ex1F	gtaaaacgacggccagtATCGTTGCAGCGTCATTGGC	577 bp
FKBP14 ex1R	caggaaacagctatgacTTTCCTGCTGGAGAGCTGGC	
FKBP14 ex2F	gtaaaacgacggccagtACTTACTGGTGGGAAAATGCAC	476 bp
FKBP14 ex2R	caggaaacagctatgacTGTGAACAGTATTAATAGACCTC	
FKBP14 ex3F	gtaaaacgacggccagtATGACAATCTTAGGAAGGCTC	532 bp
FKBP14 ex3R	caggaaacagctatgacAGATACAGTGAACCCTAAGC	
FKBP14 ex4F	gtaaaacgacggccagtCATGTCTGTGATATTGATTGGC	459 bp
FKBP14 ex4R	caggaaacagctatgacCAGAAGTCTTCTATTCAAAGACC	

Capital letters are *FKBP14* specific primers, gtaaaacgacggccagt and caggaaacagctatgac represent M13 forward and reverse primer sequences, respectively.

Table S2. **Primers used to screen for the 19 basepair deletion in exon 1 in healthy controls**

primer name	sequence (5‘- 3‘)	PCR fragment length
FKBP14 ex1del19F	TGTAGGGACTTCTTTCCTTGC	151 bp
FKBP14 ex1del19R	TGCGATGGCAGATGAATGGC	

Table S3. Primer sequences for quantitative RT-PCR

primer name	sequence (5‘- 3‘)
FKBP14-qPCR_F	ACGGCTCCTTATTTCACTCCAC
FKBP14-qPCR_R	ACACATTCCTTTCAAGCCCT
HPRT1_F	ACTTTGCTTTCCTTGGTCAG
HPRT1_R	CAAATCCAACAAAGTCTGGC