

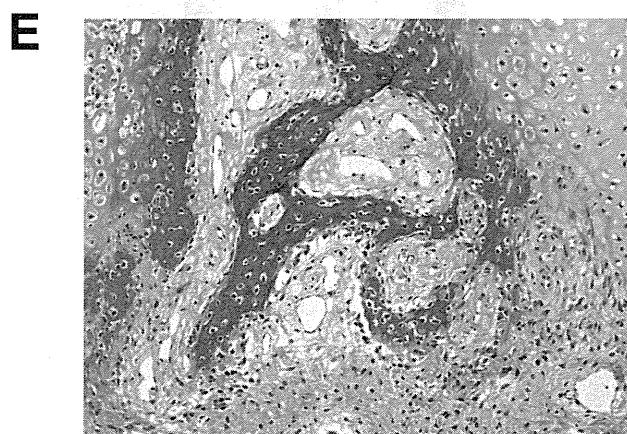
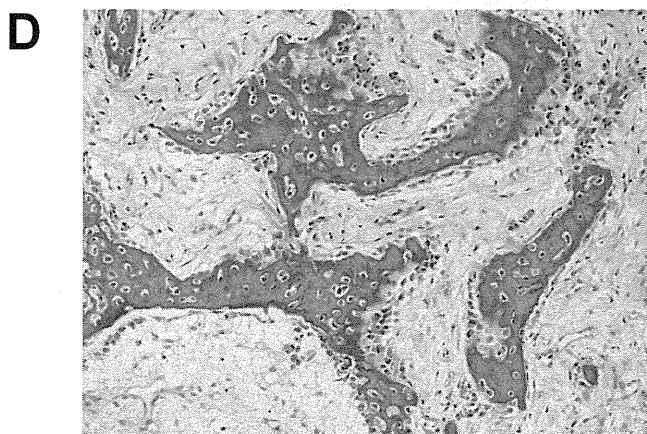
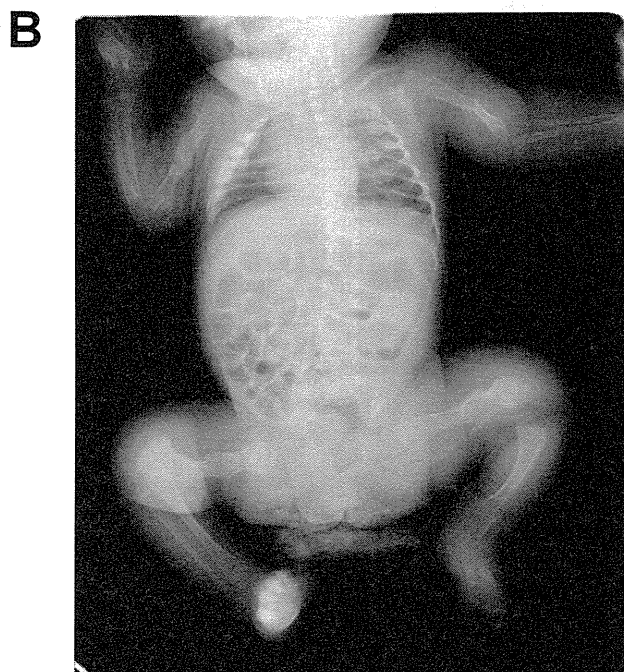
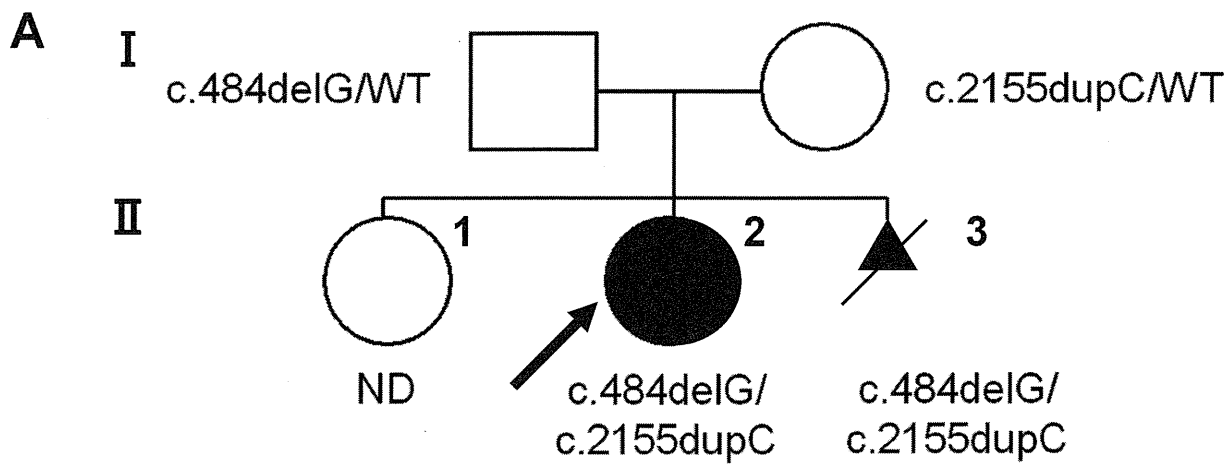
1 **FIG. 3. Characterization of the *LEPRE1* mutations and proband collagen**

2 A: Patient II-3 *LEPRE1* transcripts are about one-half the control level, by real-time RT-PCR.

3 B: Steady-state type I collagen protein from fibroblasts of Patient II-3 and a normal control is  
4 shown. In both the cell layer and media, overmodification, detected as back-streaking of collagen  
5 alpha chains ( $\alpha 1$  (I) and  $\alpha 2$  (I)) on gel electrophoresis, is present in Patient II-3. We also detected  
6 mild overmodification of type V collagen ( $\alpha 1$  (V)).

7 C: Western blots of fibroblast P3H1 in Patient II-3 and control cells confirm absence of intracellular  
8 P3H1.

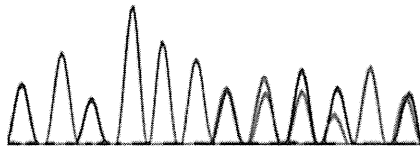
9 D: Immunofluorescent staining of fibroblasts from Patient II-3 and a normal control show  
10 colocalization of P3H1 and CRTAP with GRP94 in control cells. Both P3H1 and CRTAP proteins  
11 are absent in fibroblasts from Patient II-3.



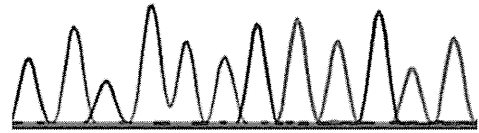
**A**

c.484delG p.A162LfsX22

Glu Lys  
G A G A A A

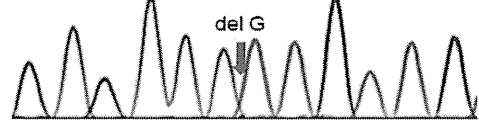


Glu Lys Ala Val  
G A G A A A G C T G T T



WT

Glu Lys Leu Leu  
G A G A A A C T G T T G



Mut

c.2155dupC p.E719RfsX11

Pro Pro  
C C C C C C

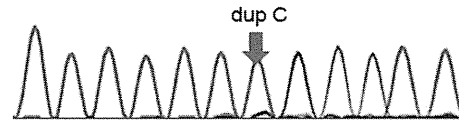


Pro Pro Glu Pro  
C C C C C C G A A C C T



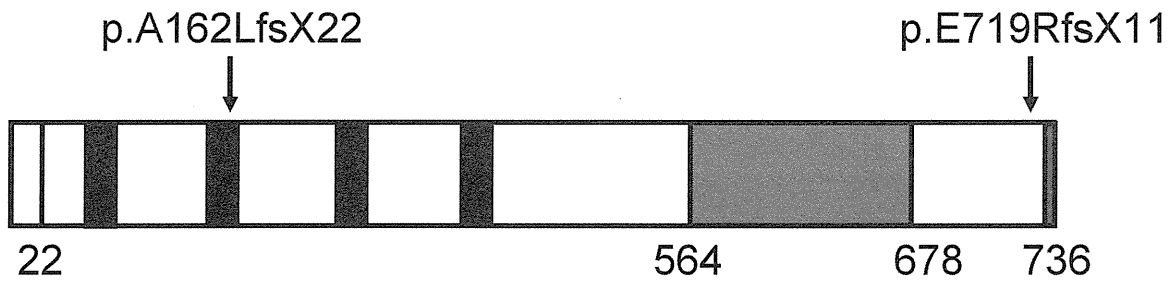
WT

Pro Pro Arg Thr  
C C C C C C G A A C C



Mut

**B**



- TPR(Tetratricopeptide repeat) domain
- PKH(Prolyl/Lysyl/hydroxylase) domain
- KDEL sequence

