

Figure 1. (A) X-rays of the skull showing generalized increase in bone density involving the skull. The sclerosis is more prominent in the base of the skull. (B) Typical end-plate thickening and sclerosis producing the classic "sandwich vertebrae" appearance. (C) Sclerosis in the iliac wings, acetabuli, and femur heads. However, typical "bone-within-bone" appearance is not notable in the adolescent. (D) The patient's father showed normal bone density, compared with the patient's characteristic sclerosis at lesions presented on the X-rays.



Figure 2. We identified a heterozygous missense mutation in the patient and his father. A heterozygous C to T transition is shown at position 746 in exon nine of *CLCN7* gene, changing a proline to leucine substitution at codon position 249.

KG sanger sequencing result
 NM_001287.5(*CLCN7*):c.746C>T, p.Pro249Leu

Depth of Coverage
 117
 % bases above 10X
 98%

