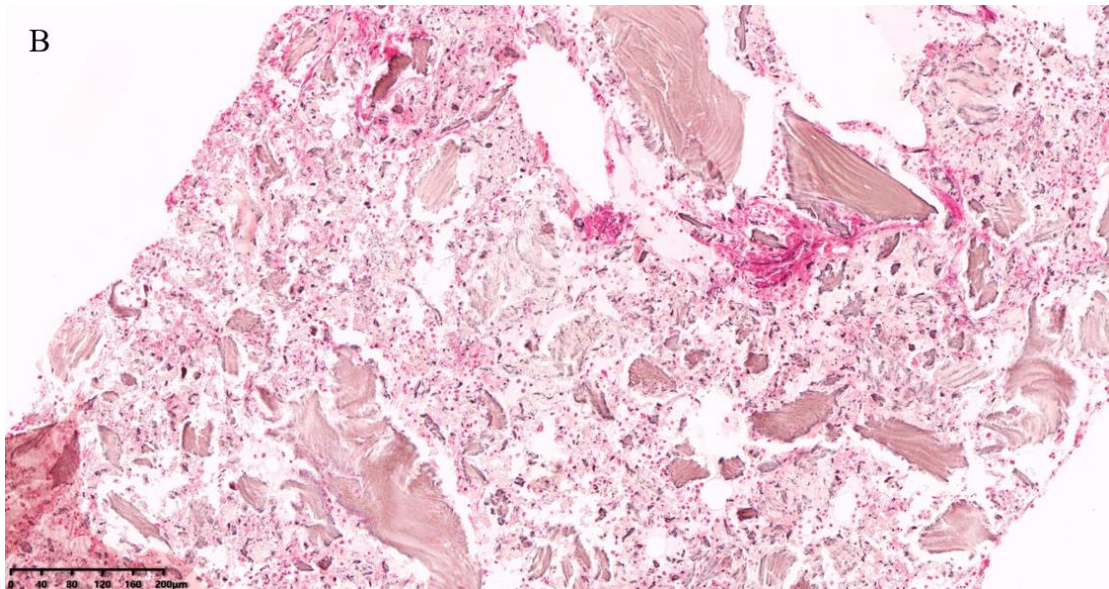
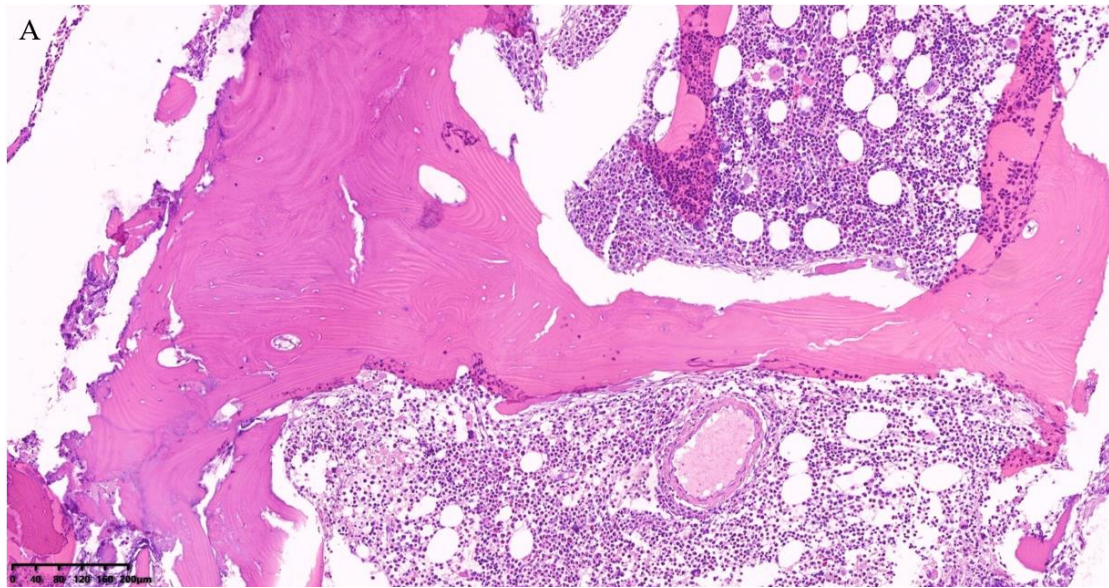


1 **Supplementary Material**



4 **Supplementary Figure 1 Bone biopsy of the proband.** A: Hematoxylin and
5 eosin staining of bone biopsy in the proband showed presence of the calcified
6 bone (pink) and active proliferation of bone marrow hematopoietic cells; B:
7 Argent– Ammonia hydroxide staining.

8

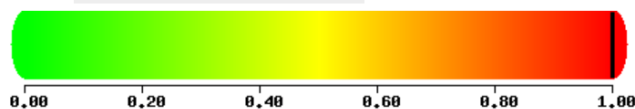
species	aa	alignment
<i>Human</i>	286	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>Mutated</i>	286	I F E Y F R R D T E K Q D F V S A G A A A G V S
<i>X.tropicalis</i>	273	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>P.troglodytes</i>	286	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>D.rerio</i>	284	M F E Y F R R D T E K R D F V S A G A A A G V S
<i>R.norvegicus</i>	284	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>M.mulatta</i>	262	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>M.musculus</i>	284	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>B.taurus</i>	290	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>G.gallus</i>	282	I F E Y F R R D T E K R D F V S A G A A A G V S
<i>C.lupus</i>	290	I F E Y F R R D T E K R D F V S A G A A A G V S

1

2 **Supplementary Figure 2 Multiple alignment of the amino acid sequence of**
3 ***CLCN7*. Multiple alignment of the amino acid sequence of *CLCN7*. R286 was**
4 **located at a highly conserved position.**

5

This mutation is predicted to be **PROBABLY DAMAGING** with a score of 1.000 (sensitivity: 0.00; specificity: 1.00)



6

7 **Supplementary Figure 3 PolyPhen-2 prediction result of the missense**
8 **mutation R286Q in *CLCN7*. Polymorphism Phenotyping v2 (PolyPhen-2) can**
9 **predict the possible impact of amino acid substitutions on the stability and**
10 **function of human proteins using structural and comparative evolutionary**
11 **considerations. The missense mutation R286Q in *CLCN7* was predicted to**
12 **have a pathogenic effect.**

13