Figure S1. Genetic testing result of the patient's father. He has the mutation in exon8 (c.1376G>A) but does not have the mutation in exon1 (c.182T>C).
Figure S2. Genetic testing result of the patient's mother. She has the mutation in exon1 (c.182T>C) but does not have the mutation in exon8 (c.1376G>A).