

Dear Reviewer 1,

We sincerely thank your valuable feedback that we have adopted to improve the quality of our paper. We have modified the manuscript to bring it to the final publication.

1. Authors should describe the background in detail why the patient has been “accidentally” (page 1) or “mistakenly” (page 2) diagnosed with ADO-II.

**Response:** We added the description of the background (page 1), and revised the core tip (page 2). About 80% of the Autosomal dominant osteopetrosis type II (ADO-II) patients were usually affected by heterozygous dominant mutation of *CLCN7* gene and presented early-onset osteoarthritis or recurrent fractures. In this study, we report the case presented with persistent joint pain without bone injury or underlying history.

2. It would be helpful if authors would provide the histologic results of pathological study to evaluate the cortical bone sclerosis and some thickened bone trabeculae.

**Response:** We added the figure (**Supplementary Figure 1**). However, the availability of the figures can provide limited value. The pathological diagnosis of this case was performed independently by a pathologist. She evaluated the cortical bone sclerosis and some thickened bone trabeculae under microscope.

3. It would be great if authors could suggest differential diagnosis except for ADO-II.

**Response:** We added the differential diagnosis in the discussion (page 8). Some differential diagnoses should be ruled out, such as congenital diseases (eg, hypoparathyroidism, pseudohypoparathyroidism), chemical poisoning

(eg, with fluoride, lead, or beryllium), and malignancies (leukemias and myeloproliferative diseases). This patient was normal urinary fluoride and PTH serum levels, and ADO-II was eventually diagnosed by analyzing radiological and genetic results.

4. While this case report described long about general information of ARO and ADO for osteopetrosis, relatively in the case, rationale arriving to the final diagnosis seem not to be well-described based on the detailed results of diagnostic work-up.

**Response:** We enriched the description of final diagnosis (page 7). Combined with the patient's medical history and radiological examination results, the final diagnosis was osteopetrosis. In the light of genetic typing (autosomal dominant form), the case belonged to ADO II.