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Autosomal Dominant Osteopetrosis type II

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Keywords:	Osteopetrosis, Osteoclast chloride channel gene mutation

Autosomal Dominant Osteopetrosis type II

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A 24-year-old previous healthy man, with no contributory family history, came to our clinic due to dyspnea and poor appetite for two weeks. In our clinics, we took a chest X-ray and a kidney, ureter and bladder (KUB) x-ray. Chest X-ray demonstrated a generalized increase of bone density (Picture 1) and vertebrae end-plate sclerotic thickening — the so-called "sandwich vertebra" appearance (Picture 2 arrow). KUB showed a typical "bone within a bone" lesion in his pelvis (Picture 3 arrow). Renal echogram revealed bilateral small kidneys without calcification or stones. Autosomal dominant osteopetrosis type II (ADOII) was diagnosed.

ADOII is a disorder with late-childhood or adult onset. Osteoclast chloride channel gene mutation (CLCN7) is responsible for the clinical manifestations. Generalized osteosclerosis developed secondary to osteoclast dysfunction. Osteosclerosis can affect the shape and structure of the bone. Some patients develop cranial nerve dysfunction or visual deficits due to osteosclerosis of skull bone and some develop bone marrow failure as a result of bone marrow cavity involvement [1, 2].

References

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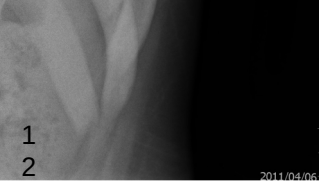

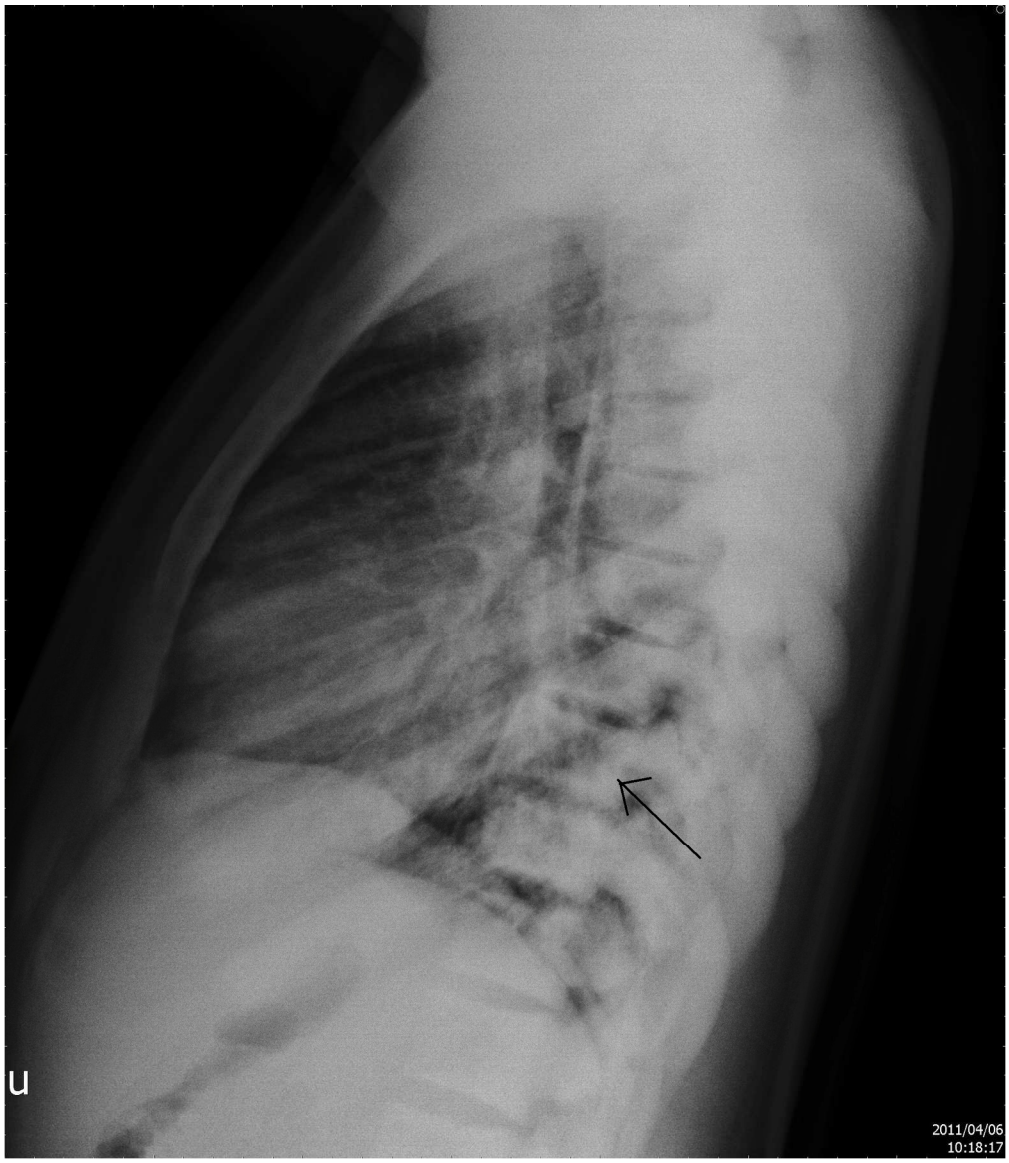

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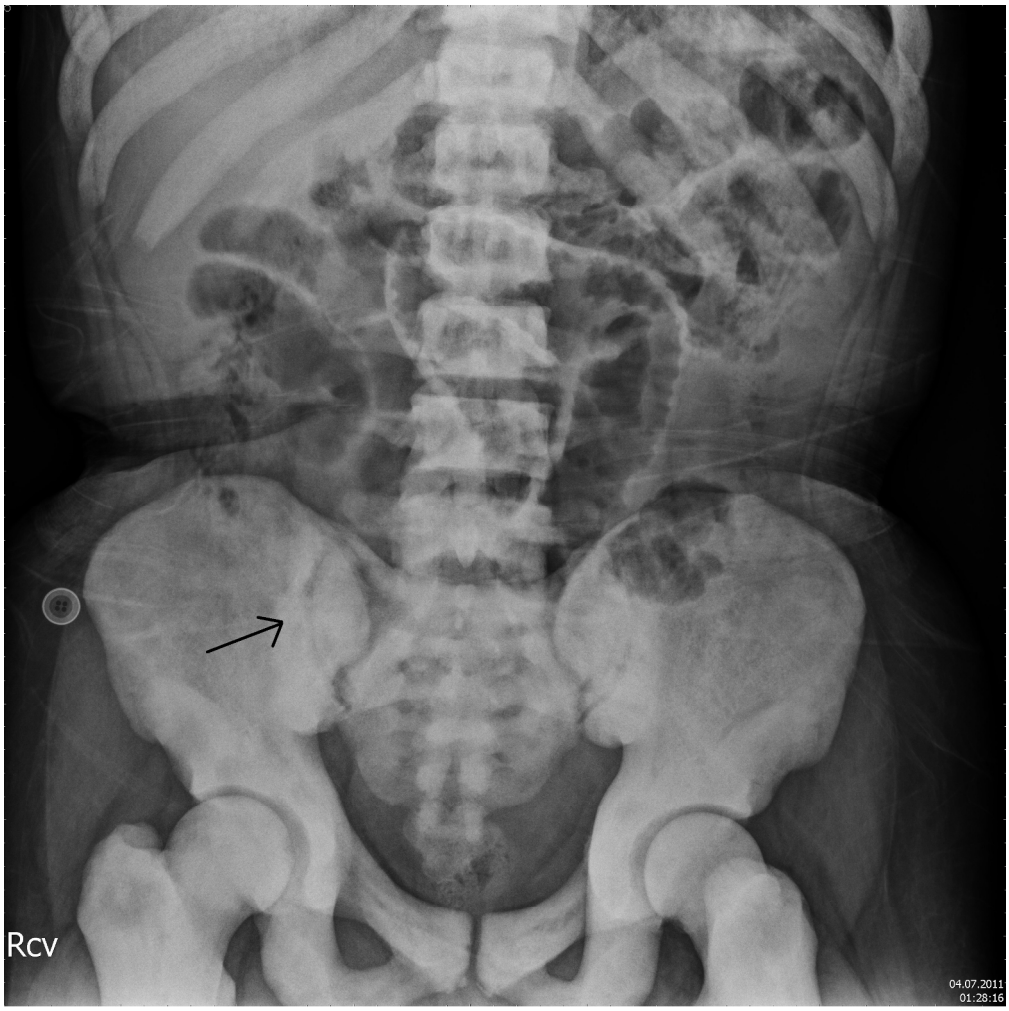

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