A unique cause of interosseous membrane calcification

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Accepted 8 January 2015

DESCRIPTION

A 55-year-old man presented with short stature, progressive bilateral leg deformity (figure 1) and loss of all teeth since childhood. Similar bony deformities were present in his mother, elder brother and both his daughters. His only son was, however, asymptomatic. There was no history of fracture. On examination, he had bilateral genu varum with an intercondylar distance of 22 cm. His biochemical evaluation was as follows (reference ranges shown parenthetically): serum calcium 2.35 mmol/L (2–2.5 mmol/L), phosphorus 0.77 mmol/L (0.81–1.4 mmol/L), alkaline phosphatase 367 mIU/mL (30–150 mIU/mL), 25(OH) vitamin D 38 nmol/L (75–250 nmol/L), parathormone 11.2 pmol/L (0.9–5.5 pmol/L) and creatinine 71 μmol/L (54–106 μmol/L). His tubular maximum phosphate/glomerular filtration rate was 0.88 mmol/L (1.12–1.8). X-ray of the forearm showed calcification of the interosseous membrane (figure 2). Dual-energy X-ray absorptiometry scan revealed osteoporosis at his left hip (T score of −2.6) with paradoxically increased bone density at lumbar spine (T score of 2.8). Biochemical evaluation of his mother, elder brother and his daughters showed hypophosphatemia. The differential diagnosis of interosseous membrane calcification includes fluorosis,1 X linked hypophosphatemic rickets and osteogenesis imperfecta type V.2 The presence of three generational family history, bony deformity with dental anomalies, persistent hypophosphatemia and radiological evidence of enthesopathy pointed to a probable diagnosis of X linked hypophosphatemic rickets. Enthesopathy is a well-documented complication of X linked hypophosphatemic rickets. This explains paradoxical increase in bone density at lumbar spine in our case. Calcification of interosseous membrane is a rare manifestation of enthesopathy in X linked hypophosphatemic rickets.3

In less-developed countries, bony and dental deformity with familial or geographic clustering may also occur in fluorosis. The absence of fracture serves to exclude osteogenesis imperfecta as possible cause. The biochemical hypophosphatemia in the patient’s family, with characteristic vertical genetic transmission without skipping of generations, absence of father to son transmission and occurrence in all daughters of an affected father, all serve to diagnose the characteristic X linked dominant inheritance in patient with hypophosphatemic rickets.

Learning points

▸ The differential diagnosis of interosseous membrane calcification are: X linked hypophosphataemic rickets, fluorosis and osteogenesis imperfecta type V.
▸ Presence of hypophosphatemia rules out fluorosis and osteogenesis imperfecta type V.
▸ Absence of father to son transmission in the family strengthens the diagnosis.

Contributors RK wrote the article and collected the data. KB modified the content and edited the manuscript. SK provided the images and modified the content. JPS made the diagnosis.
REFERENCES

