Neurofibromatosis, Pathological Fracture and Hypervitaminosis-D

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Neurofibromatosis may rarely present with pathological fracture(1). We present such a child who was misdiagnosed as a case of rickets and treated repeatedly with vitamin D injection resulting in iatrogenic hypervitaminosis D.

CASE REPORT

A four and half years old female child was referred for management of bowing of left leg and pathological fracture. This was associated with vomiting, abdominal pain, increased thirst and increased volume of urine. There was no history of joint pain, convulsion and difficulty of vision. The child was a product of a non-consanguineous marriage. She was of average weight at birth and did not have any neonatal problem. Her mother was having multiple cuteneous neurofibromas and was diagnosed as a case of neurofibromatosis (NF) I.

Her developmental history revealed mild delay in motor development. The immunization status and dietary intake was normal. She was diagnosed as suffering from rickets by different physicians at different places; many of whom treated her with injection arachitol (Vitamin D3). Overall, she received about nine such injections over a period of three months. Then she developed features of hypercalcemia with polyuria, polydipsia, nausea, vomiting and abdominal pain.

Her anthropometry revealed height of 96 cm (<5th percentile), weight of 10 kg (<5th percentile) and head circumference of 47 cm. She had multiple café au lait spots over the trunk and extremities and bowing of left leg without any features of rickets. Her systemic and ocular examination was within normal limit.

Her hematological profile, renal function, liver function and routine urine and stool examination were within normal limits. Chest X-ray was also normal. X-ray spine showed normal outline of lumbar vertebrae with normal disc space. Serum ionized calcium was 6.9 mEq/L (normal-2.3-5.2 mEq/L), alkaline phosphatase 380 IU/L (N 145-420 IU/L), inorganic phosphate was 2.0 mEq/L (2.3-3.5 mEq/L), and serum parathormone level was 3.15 pg/mL(15-65 pg/mL). X-ray lower limb showed bowing and pathological fracture of left fibula.

DISCUSSION

Our patient met three of the seven diagnostic criteria of neurofibromatosis. Our patient developed a pathological fracture in the left fibula that was initially misdiagnosed as rickets. She was injected with vitamin D3, which resulted in hypercalcemia and polyuria. The radiological findings observed were consistent with pathological fracture of the left fibula. The management of such cases should include a comprehensive approach involving a multidisciplinary team approach.
criteria(2): presence of multiple café’u lait spots, an osseous lesion as bony dysplasia in the form of bowing of legs and cortical thinning (which had lead to pathological fracture) and mother suffering from Neurofibromatosis (NF) I. She had been wrongly diagnosed as a case of pathological fracture due to rickets from elsewhere and inadvertently treated with repeated doses of vitamin D3 injections by various local physicians causing hypervitaminosis D that manifested as hypercalcemia, hypophosphate-mia and low serum parathormone level.

Apart from bowing of tibia and fibula, leading to pathologic fracture, many other skeletal abnormalities like kyphoscoliosis, dysplasia of sphenoid wing, pseudoarthrosis, non-ossifying fibroma etc, may be associated with NF(3,4). Treatment should be aimed at preventing pathological fractures. Pseudarthrosis should be managed with corrective operation or osteotomy as appropriate(5).

Our patient did not have any clinical or biochemical features of rickets. Rickets or osteomalacia may also occur in neurofibromatosis, though rarely. The exact pathogenetic mechanism for this remains to be determined, but it may be due to phosphatonin mediated inhibition of renal tubular reabsorption of phosphate and reduced serum phosphorus. Characteristically, it has a later onset in adulthood. Rickets in NF typically has multiple pseudo fractures and responds to treatment with pharmacological dose of vitamin D with or without phosphate supplementation(6).

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REFERENCES


