CASE REPORT

Neurofibromatosis, Pathological Fracture and Hypervitaminosis-D

RAKESH MONDAL, MADHUMITA NANDI AND PK CHANDRA

From the Department of Pediatric Medicine, IPGMER and SSKM Hospital, Kolkata, West Bengal, India.

Corrspondence to: Dr Madhumita Nandi, 6/6, Naren Sarkar Road, Barisha, Kolkata, West Bengal 700 008, India.
madhumitabanik@ rediffmail.com
Received: April 15, 2009;
Initial review: May 13, 2009;
Accepted: July 7, 2009.

Pathologic fractures in children may be due to various causes. Rarely, it may be the presenting symptom of neurofibromatosis. A misdiagnosis of Rickets and Vitamin D supplementation in such a case may wreak havoc in the form of iatrogenic hypervitaminosis D. We report one such case.

Key words: *Hypervitaminosis D, Neurofibromatosis, Pathologic fractures.*

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(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, pseudoarthosis, non-ossifying fibroma etc, may be associated with NF(3,4). Treatment should be aimed at preventing pathological fractures. Pseudar-throsis 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 pseudofractures and responds to treatment with pharmacological dose of vitamin D with or without phosphate supplementation(6).

Contributors: All three authors were involved in diagnosing and managing the case. RM drafted the manuscript. MN and PKC searched the literature. All three approved the final manuscript.

Funding: None.

Competing interests: None stated.

REFERENCES

1. Arroba Basanta ML, de la Rubia Fernandez LR, del Castillo Martin F. Neonatal pathologic fracture and neurofibromatosis. An Esp Pediatr 1986; 25:491-492.
2. Haslam RHA. Neurocutaneous syndrome. *In*: Behrman RE, Kilegman RM, Jenson HB, editors. Nelson Textbook of Pediatrics. 18th edition. Philadelphia: WB Saunders Company; 2008. p. 2483-2484.
3. Hofmann P, Galanski M. Congenital bowing of the tibia in neurofibromatosis von Recklinghausen (author's transl). Fortschr Geb Rontgenstr Nuklearmed 1976; 125: 417-421.
4. Howlett DC, Farrugia MM, Ferner RE, Rankin SC. Multiple lower limb non-ossifying fibromas in siblings with neurofibromatosis. Eur J Radiol 1998; 26 : 280-283.
5. Ramelli GP, Slongo T, Tschappeler H, Weis J. Congenital pseudarthrosis of the ulna and radius in two cases of neurofibromatosis type 1. Pediatr Surg Int 2001; 17: 239-241.
6. Konishi K, Nakamura M, Yamakawa H, Suzuki H, Saruta T, Hanaoka H, *et al*. Hypophosphatemic osteomalacia in von Recklinghausen neurofibromatosis. Am J Med Sci 1991; 301: 322-328.