Bilateral Transverse (Bowdler) Fibular Spurs with Hypophosphatasia in an Adolescent Girl

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Hypophosphatasia is a clinically heterogeneous inheritable disorder characterized by defective bone mineralization and the deficiency of serum and tissue liver/bone/kidney alkaline phosphatase activities. Due to the mineralization defect of the bones, various skeletal findings can be radiologically observed in hypophosphatasia. Bowing and Bowdler spurs of long bones are the characteristic findings. The Bowdler spurs reported on in the previous pertinent literature were observed in the perinatal aged patients and these lesions have rarely involved adolescents. We herein report on a 14-year-old girl with fibular Bowdler spurs.

Index terms:
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Hypophosphatasia is a rare inheritable inborn error of metabolism that is characterized by defective bone mineralization and deficiency of the serum and tissue liver/bone/kidney alkaline phosphatase activities (1). The patients exhibit considerable skeletal pathologies such as bowing deformities, shortening of bones, cortical thickening and pseudofractures (2). The relevant literature mainly reports on perinatal Bowdler spurs; thus, in this report, we present an adolescent hypophosphatasia patient with congenital fibular Bowdler spurs.

CASE REPORT

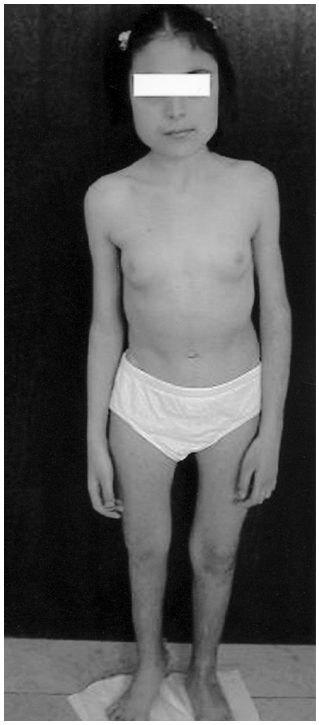
A 14-year-old girl was referred to our clinic because of her growth retardation and scoliosis. She had been born via caesarian section at 38 weeks with a birth weight of 1,500 grams, and she had been observed in the neonatal intensive care unit for three days. Her Bowdler spurs had been first noticed as ossifications extending into the skin. Her laboratory values obtained at the second day of birth were as follows: serum alkaline phosphatase 8 IU (reference range 430–1,140 IU), serum calcium 2.30 mmol/L and serum phosphate 1.82 mmol/L. The patient had been diagnosed as having hypophosphatasia and so she had been followed up by the pediatric endocrinology department. The patient had been lost to follow-up by her own choice and had not received any treatment for 12 years.

Her family history was insignificant except for the congenital hip dysplasia of her mother and the growth retardation of her 13 year old brother, and her 12 year old sister was healthy. Her parents were non-consanguineous.

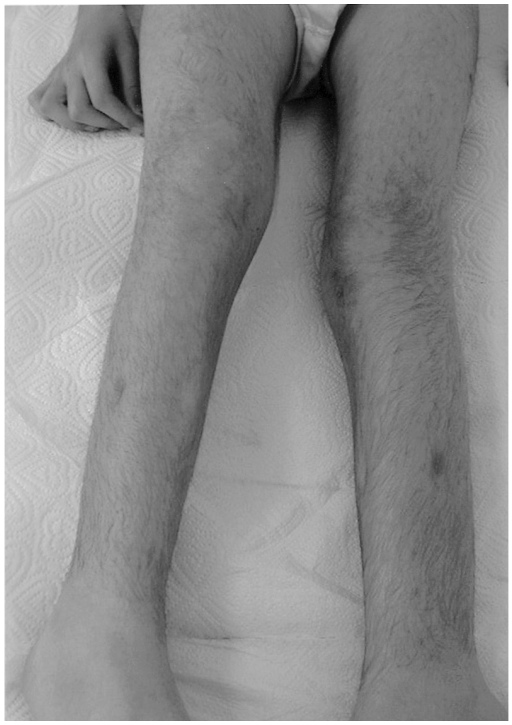
The physical examination revealed marked growth retardation (weight 36 Kg [< 5 percentile], height 126 cm [< 5 percentile]). Her right lower extremity was 2 cm shorter than her left leg. Her right femur displayed bowing and an external rotation deformity with limitation of internal rotation. The skin overlying the Bowdler spurs showed fibrotic dimpling and brown discoloration (Figs. 1A, B). She had bilateral pes

planovalgus (Figs. 1A, B). Her teeth development was normal. The laboratory values were as follows: serum calcium: 9.7 mg/dl, alkaline phosphatase 12 IU and phosphorus 6 mg/dl.

Radiological examination showed bowing of the ribs, lucent streaks in the metaphysis representing nests of unossified physeal cartilage, and bowing and widening of the femur. The Bowdler spurs in both fibulae and in the right hip joint femoral head were directed anteriorly and superiorly (Fig. 1C). The femoral necks were short and



A



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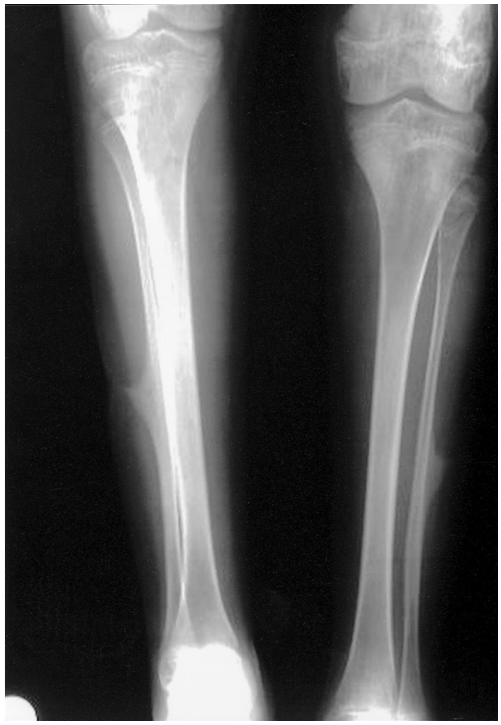
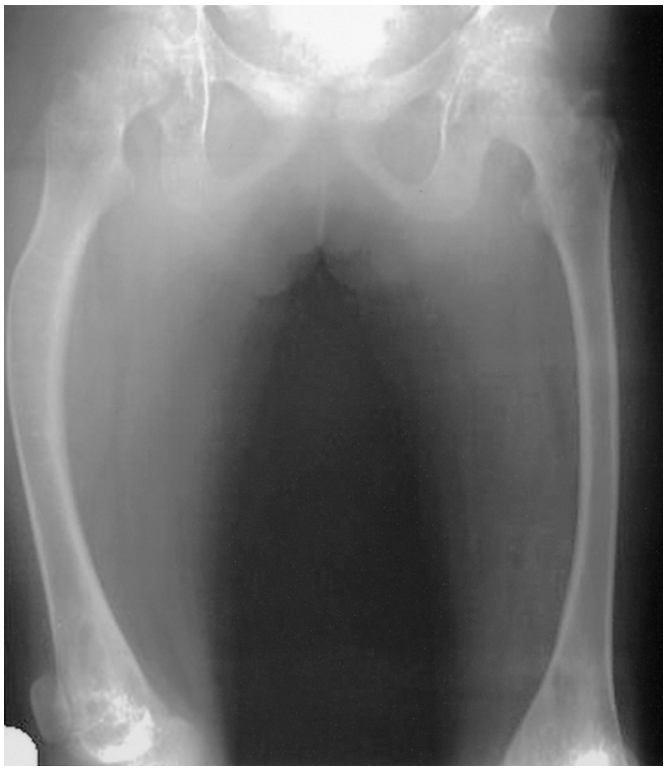


C



D

Fig. 1. A 14-year-old girl with hypophosphatasia.
A. The general appearance of the patient with marked growth retardation and shortness in her right lower extremity.
B. The skin overlying the Bowdler spurs shows fibrotic dimpling and brown discoloration with bilateral pes planovalgus deformity of the feet.
C. Bilateral fibular Bowdler spurs.
D. An irregularity of the articular surfaces of both the femoral head and the acetabulums with narrowing of the joint space, and the diffuse osteoporosis and radiolucent areas in all the bones are prominent.

wide, and there was an increase of the collodiaphyseal angle with anterosuperior displacement of the femoral heads (Fig. 1D). There was irregularity at the articular surface of both femoral heads and the acetabulums with narrowing of the joint spaces. There was diffuse osteoporosis and radioluscent areas in all the bones (Figs. 1C, D).

DISCUSSION

Hypophosphatasia is a rare inheritable inborn error of metabolism that was first described by Rathbun (3). This disorder is characterized by defective bone mineralization and a deficiency of serum and liver/bone/kidney tissue alkaline phosphatase activities (1). Deficiency of alkaline phosphatase results in the accumulation of inorganic pyrophosphates, and this results in undermineralized bone with incomplete ossification of the cartilage and the metaphyseal regions (2). Large amounts of unmineralized osteoid are present in all bones, and this is especially seen in the metaphyseal regions (2).

Hypophosphatasia causes defective skeletal mineralization that manifests clinically as rickets in infants and children, and it manifests as osteomalacia in adults. Six clinical forms are currently recognized: perinatal, infantile, childhood, adult, odontohypophosphatasia and a benign prenatal form that was decently described by Pauli et al. (4). When hypophosphatasia presents clinically in a severe form either before or during the newborn period, perinatal death occurs. By contrast, the infantile type presents during the first six months of life with growth failure and rachitic like skeletal deformities that result in recurrent respiratory infections and increased intracranial pressure (5). Our patient had Bowdler spurs at birth, but she did not have any serious disease till she reached 14 years old. Accordingly, we believe that our patient had the benign prenatal form of hypophosphatasia.

A wide spectrum of the radiographic and clinical abnormalities seen in perinatal hypophosphatasia was presented by Kozlowski and colleagues (6). Ossification is generally compromised in the entire skeleton and a complete lack of ossification ensues in some individual bones. Significant radiological changes are known to ensue with hypophosphatasia, and they include very thin and wavy fractures of the ribs, lucent defects and irregularities, "metaphyseal cupping", "chrosomal like" or "campomelic like" changes of the femur and humerus, various decreases of ossification in the radius, ulna, tibia and fibula, and the characteristic "spurs" of the ulna and fibula (at the midshaft) that protrude towards the skin (6).

In the literature, the Bowdler spurs that have been reported on up to now are generally in the perinatal age group. We have presented here a rare adolescent case of hypophosphatasia with Bowdler spurs; thus, we advocate that the diagnosis of hypophosphatasia also be considered for those adolescent patients having radiological evidence of Bowdler spurs.

References

1. Whyte MP. Hypophosphatasia and the role of alkaline phosphatase in skeletal mineralization. *Endocr Rev* 1994;15: 439-461
2. States LJ. Imaging of metabolic bone disease and marrow disorders in children. *Radiol Clin North Am* 2001;39:749-772
3. Rathbun J. Hypophosphatasia, a new developmental anomaly. *Am J Dis Child* 1948;75:822-827
4. Pauli RM, Modaff P, Sipes SL, Whyte MP. Mild hypophosphatasia mimicking severe osteogenesis imperfecta in utero: bent but not broken. *Am J Med Genet* 1999;86:434-438
5. Whyte MP, Vrabel LA, Schwartz TD. Alkaline phosphatase deficiency in cultured skin fibroblasts from patients with hypophosphatasia: comparison of the infantile, childhood, and adult forms. *J Clin Endocrinol Metab* 1983;57:831-837
6. Kozlowski K, Sutcliffe J, Barylak A, Harrington G, Kemperdick H, Nolte K, et al. Hypophosphatasia. Review of 24 cases. *Pediatr Radiol* 1976;5:103-117