A case of osteopetrosis with pelvic ectopic spleen: an unusual association

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SUMMARY: Reisli İ, Çalışkan Ü, Taştekin G, Koç H, Açıkgözoğlu S, Aydoğdu-Kıreşi D, Aydın K. A case of osteopetrosis with pelvic ectopic spleen: an unusual association. Turk J Pediatr 2001; 43: 265-268.

A three-month-old girl was admitted to the hospital with a history of pallor. On physical examination, the liver was enlarged and a solid mass was palpated in the left abdomen. Laboratory evaluation revealed anemia and thrombocytopenia. Bone marrow was hypocellular with reduced number of megakaryocytes. Radiographic findings and scintigraphic study of the long bones were consistant with osteopetrosis. In the imaging studies, including ultrasonography, computerized tomography, magnetic resonance imaging and scintigraphic study, an ectopic spleen expanded into the bony pelvis was observed. We report here unique case of infantile osteopetrosis associated with pelvic ectopic spleen.

Key words: infantile osteopetrosis, ectopic spleen, hypersplenism.

Osteopetrosis is a rare bone disease characterized by a generalized increase in the density of the skeleton. Because of the defective osteoclast function, normal bone growth and remodeling are impaired. Several forms of the disorder, with different prognoses and modes of genetic transmission, are now recognized[1-3]. Another very rare entity is pelvic ectopic spleen. It occurs as a consequence of the embryonal disturbances in the development of the ligaments connecting the spleen with surrounding tissue[4,5]. We report a case of infantile osteopetrosis associated with pelvic ectopic spleen.

Case Report

A three-month-old girl was admitted to the hospital with a history of pallor and irritability. Her family history was only significant for consanquinity. On her physical examination, her height and head circumference were below the 3rd percentile, the liver was enlarged, and a solid mass about 13 x 7 cm in diameter, expanding from 1 cm below the left costal margin toward the right pelvic region, was palpated.

Her laboratory evaluation revealed hemoglobin 51 g/L, white blood cell count 11.8 x 10^9/L and platelet count 31 x 10^9/L. A peripheral blood smear showed normocytic, mild hypochromic erythrocytes; mild anisopoikilocytosis; basophilic stippling; polychromasia; and numerous myeloid progenitors. The reticulocyte count was 7.1%. Bone marrow aspiration was performed with difficulty and revealed hypocellular, markedly reduced megakaryocytes, and no abnormal cells. The HbA was 93%, HbF 6.2%, and HbA_2 0.8% in the hemoglobin electrophoresis. The results of the serological tests for Toxoplasma, cytomegalovirus, herpes, and syphilis were negative. The level of alkaline phosphatase was increased (1164 U/L), but the other laboratory measurements were normal.

In radiographs of the cranium, chest, pelvis and long bones, there was a generalized increase in bone density, and the demarcation between the cortex and the medullary cavity was lost (Fig. 1). In the scintigraphic scan of the bone [3 mCi Tc-99m methylenediphosphanate (MDP); DUPONT PHARMA], there was generalized intense uptake of radiophosphate in the skeleton with faint visulization of the kidneys (Fig. 2).

In the direct radiographs, there was no spleen shadow in the area where it is normally seen, and in the pelvic region, there was a soft tissue

density pushing the intestines above. In the abdominal ultrasonography, an enlarged liver was observed and the spleen was again not observed in its region; however, a solid mass expanding into the bony pelvis was found in the lower region of the abdomen. In the Doppler ultrasonography, the spleen's arterial blood supply was from the celiac plexus, and its venous outflow was into the portal vein. In computerized tomography, there was an ectopic spleen. This ectopic spleen with its vascular structure was also confirmed by abdominal magnetic resonance imaging (Figs. 3a and 3b). In the scintigraphic examination (2 mCi Tc-99m tin colloid; DUPONT PHARMA), there was an ectopic spleen showing diffuse hyperplasia in the pelvic region (Fig. 4). High-dose methylprednisolone was used for the treatment of anemia and thrombocytopenia, but the patient died on the 13th day of treatment from pulmonary infection.

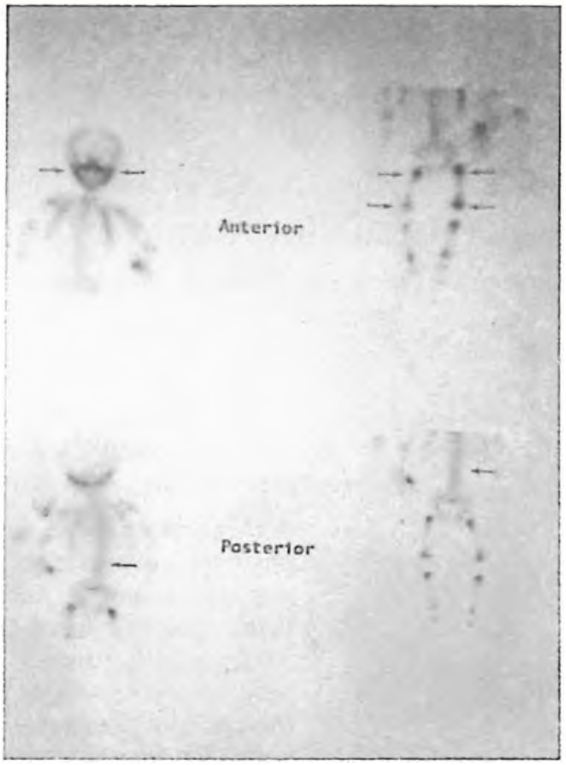

Fig. 2. Scintigraphic study of the bone: Tc-99m MDP scan shows a generalized intense radiophosphate accumulation in the skeleton (anterior) with faint visualization of the kidneys (posterior).

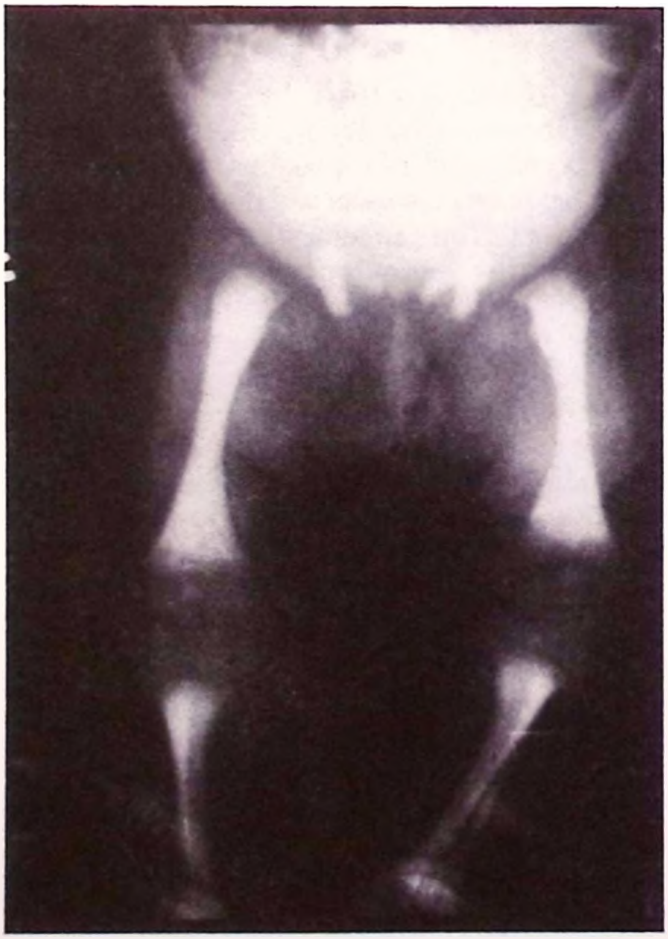

Fig. 1. Generalized increased density of the long bones and loss of demarcation between the cortex and medulla.

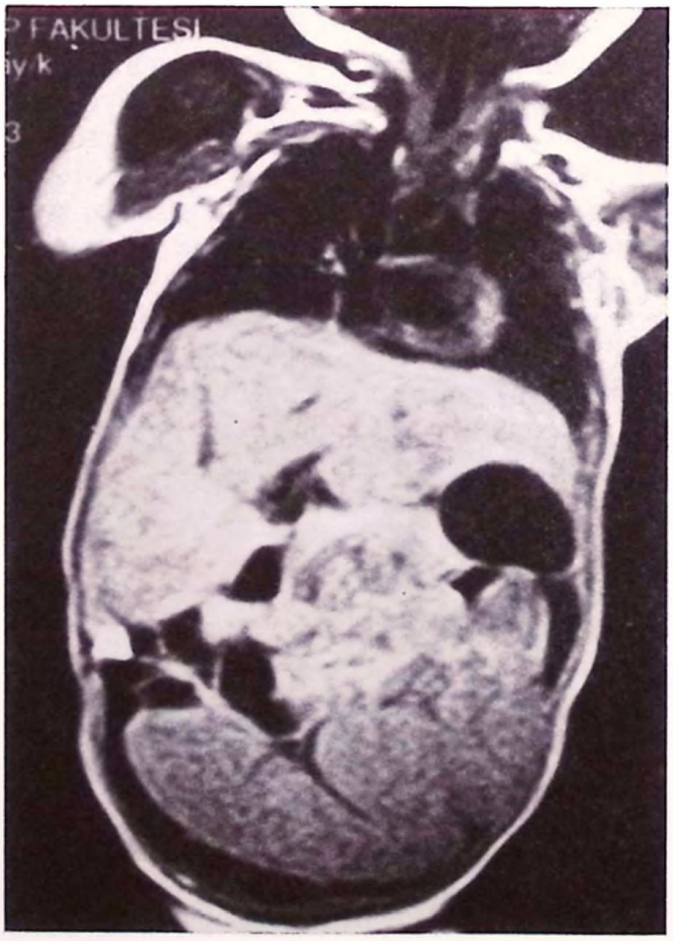

Fig. 3a. Pelvic ectopic spleen and its vascular structure in magnetic resonance imaging (coronal section).

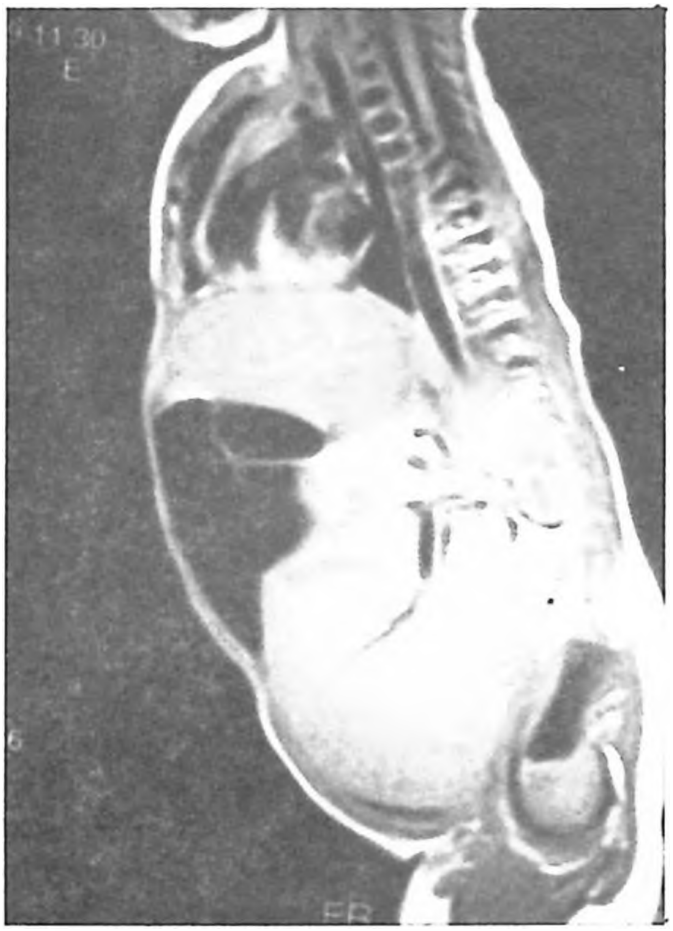

Fig. 3b. Pelvic ectopic spleen and its vascular structure in magnetic resonance imaging (sagittal section).

Fig. 4. Pelvic ectopic spleen in scintigraphic study.

Discussion

Osteopetrosis appears with mild or severe clinical forms. The mild type was originally described in 1904 by Albers-Schönberg with autosomal dominant inheritance. Conversely, the autosomal recessive type of osteopetrosis is the most severe, and is termed the "malignant" form. In addition to these two forms, an autosomal recessive form of intermediate severity is recognized[2,3]. The autosomal dominant type is seen in older children and adults. It is often asymptomatic, but may be associated with pathologic fractures, mild anemia and cranial nerve paralysis. The autosomal recessive form of osteopetrosis, or infantile osteopetrosis, typically appears in infancy and the patients die in early childhood. The clinical manifestations of infantile osteopetrosis include failure to thrive, hepatosplenomegaly, severe anemia and cranial nerve dysfunction. The volume of the bone marrow is reduced by progressive obliteration of the marrow space by the increased mass of sclerotic bone, which makes bone marrow aspiration difficult. Hypersplenism eventually develops and leads to pancytopenia[1-3]. Our patient was accepted as infantile osteopetrosis with hypersplenism based on the severe clinical manifestations in early infancy and laboratory findings.

A pelvic ectopic spleen was another interesting finding in our patient. It is an extremely rare entity and should be considered in the differential diagnosis of abdominopelvic masses. Radiological and scintigraphic studies can be used to locate and identify these conditions[4-7]. After ultrasonography, computerized tomography, magnetic resonance imaging and scintigraphic study, we determined the localization, vascularization and function of the pelvic mass and confirmed the diagnosis of pelvic ectopic spleen.

The association of infantile osteopetrosis and pelvic ectopic spleen has not been reported in the literature prior to this case report.

REFERENCES

1. Yavuz H, Çalışkan U, Bozkır A, Arslan A, Koç H, Wood BP. Radiological cases of the month: autosomal recessive osteopetrosis. Am J Dis Child 1992; 146: 1309-1310.

2. Elster AD, Theros EG, Key LL, Stanton C. Autosomal recessive osteopetrosis: bone marrow imaging. Radiology 1992; 182: 507-514.

3. Alter BP, Young NF. Bone marrow failure syndromes. In: Nathan DG, Orkin SH (eds). Nathan and Oski's Hematology of Infancy and Childhood (5th ed). Philadelphia: WB Saunders; 1998: 237-335.
4. Popovic M, Kenezevic S, Colovic R, Petrovic M, Antunovic P, Djordejvic Z. [Ectopic spleen-2 case reports.] Acta Chir Iugosl 1995; 42: 59-62.
5. Azar GB, Awward JT, Mufarrij IK. Accessory spleen presenting as adnexal mass. Acta Obstet Gynecol Scand 1993; 72: 587-588.
6. Lamesch P, Lamesch A. Anomalies of the position of the spleen in the child. Case report and review of the literature from 1896 to 1990. Langenbecks Arch Chir 1993; 378. 171-177.
7. Cendrowski K, Savicki W, Stelmachow J, Wypych K. Ectopic spleen in pelvis minor-a case report. Ginekol Pol 1992; 63: 312-314.