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INFANTILE MYOFIBROMATOSIS*

A Case Report

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Infantile myofibromatosis is a rare mesenchymal disorder of infancy characterized by the formation of tumors in the skin, muscle, viscera, bone and subcutaneous tissue. The etiology of the disorder is unknown. We describe here a newborn with multiple infantile myofibromatosis, peritonitis and intestinal perforation. Surgery revealed multiple intestinal obstructions and jejunal perforation due to intestinal tumors; consequently, a jejunostomy was performed. The patient was maintained on total parenteral nutrition and oral semiliquid infant formula for two months, however he died due to multiple attacks of diarrhea and septicemia. *Key words: infantile myofibromatosis, newborn intestinal obstruction, intestinal perforation.*

Infantile myofibromatosis (IM) is a rare mesenchymal disorder in newborns and infants, and is characterized by the formation of tumors involving the skin, skeletal muscle, viscera, bone and subcutaneous tissues[1,2]. It was first documented in 1954 and called "congenital generalized fibromatosis"[2,3]. It is also known as generalized hamartomatosis, multiple congenital mesenchymal tumor, diffuse congenital fibromatosis, and multiple vascular leiomyomas of the newborn[1,2]. Chung and Enzinger[4] named the entity "infantile myofibromatosis" in 1981.

We describe here a newborn with multiple infantile myofibromatosis involving the gastrointestinal tract, which was perforated as a result.

Case Report

A four-day-old boy was admitted to the Department of Pediatric Surgery, Faculty of Medicine, Trabzon, with bilious vomiting, abdominal distension and multiple subcutaneous nodules. The infant was the product of a full-term pregnancy ending in spontaneous delivery with no apparent complication. The mother had no prenatal care and there was no family history of disease.

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On physical examination, the infant was found to have multiple subcutaneous nodules, abdominal distension, dehydration and angulation of the right thigh. The subcutaneous nodules were firm, mobile and 0.5 to 4.0 cm in diameter in the scalp, thoracal wall and extremities (Fig. 1). Abdominal plain films showed free air and air fluid levels. Plain films of the body demonstrated osteolytic lesions of the extremities and a fracture of the right femur (Fig. 2).

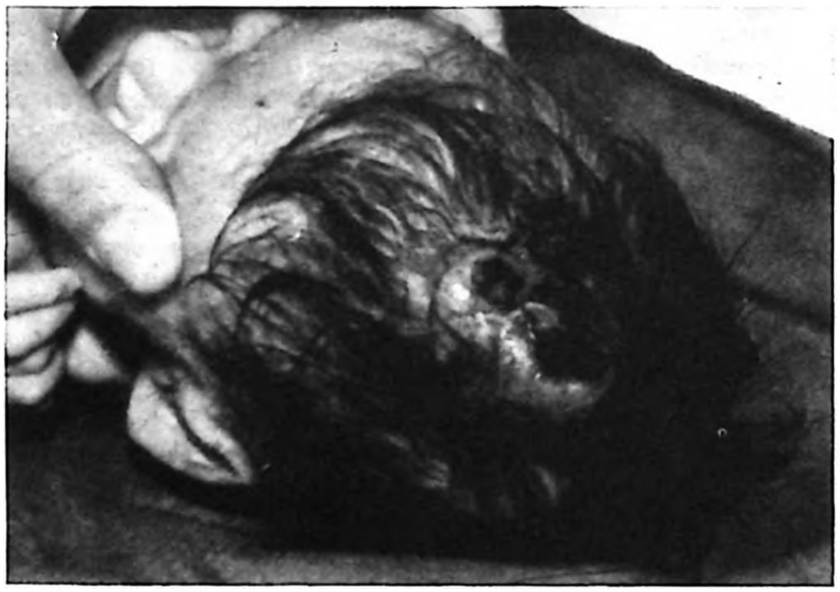

Fig. 1: The mass in the parietal region is 4 cm in diameter. The overlying skin was ulcerated.

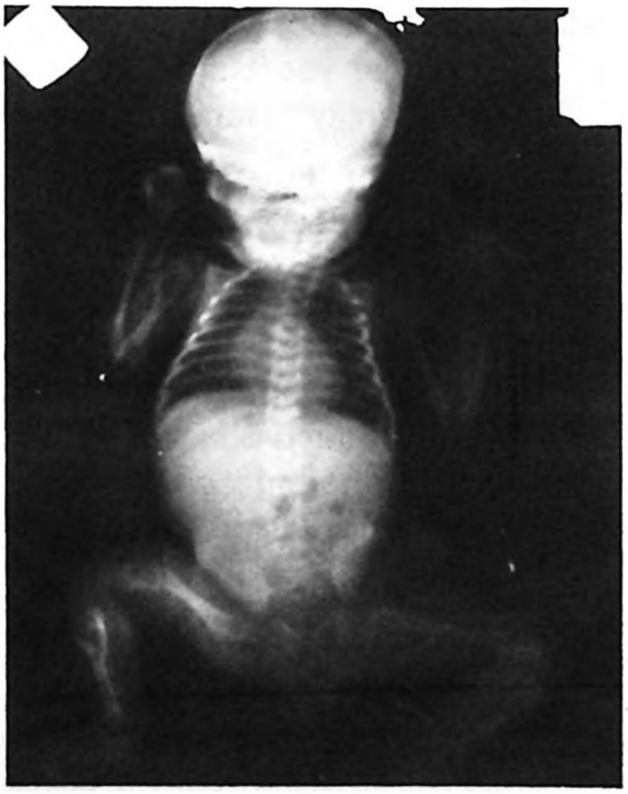

Fig. 2: Whole-body, post-operative x-ray showing osteolytic lesions of the extremities and fracture of the right femur.

After resuscitation with intravenous fluid, injection of vitamin K, nasogastric suction and antibiotics, an emergency laparotomy was performed through a right, transverse, supraumbilical incision. Peritonitis, multiple nodules from the stomach to the sigmoid colon and parietal peritoneum, and jejunal perforation 40 cm distal to the ligament of Trietz in the intact intestine were found. In addition, there were multiple intestinal obstructions in the jejunum, ileum and colon due to tumors. After washing the peritoneal cavity with hot saline, a biopsy specimen from the jejunal segment was obtained for pathological examination, a jejunostomy was performed and the abdominal wound was closed in a single layer. A subcutaneous nodule was excised for pathologic examination. Subsequently, traction of the right leg was performed for the femur fracture.

Parenteral nutrition was started postoperatively, and following the removal of the nasogastric tube on the fifth day, liquid oral nutrient was started. Histologic examination of subcutaneous and intestinal nodules indicated features of infantile myofibromatosis (Fig. 3). On the ninth postoperative day, semiliquid infant formula and colestyramine were permitted.

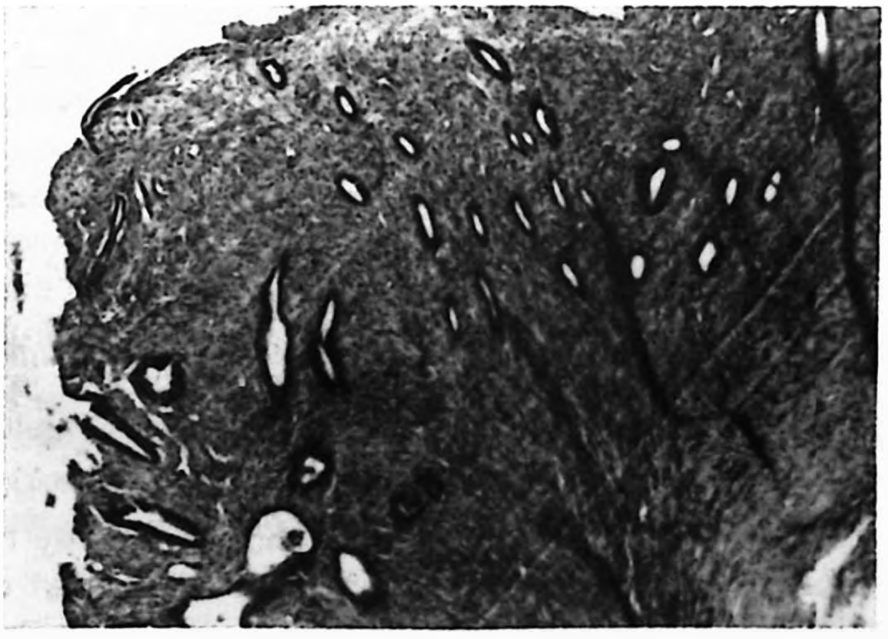

Fig. 3: Photomicrograph from the intestinal lesion demonstrating the characteristic appearance of infantile myofibromatosis.

In the postoperative period, the patient developed multiple attacks of severe diarrhea. Two months later, the infant's general status deteriorated and respiratory distress developed. After two days the infant died due to septicemia. The blood culture grew Pseudomonas aeruginosa resistant to all antibiotics.

Discussion

Infantile myofibromatosis is a rare mesenchymal disorder manifested by the formation of tumors in the skin, muscle, viscera, bone and subcutaneous tissues. The etiology of the disorder is uncertain. There is evidence to support that infantile myofibromatosis is a heritable condition[3-5]. It has also been suggested that IM lesions are hamartomatous in nature[6], and that IM results from in utero stimulation by estrogenic hormones[2, 6, 7].

It has been well established that there are two types of IM: i) the solitary lesion, and ii) the multiple lesion[1, 2]. The solitary lesion usually involves the skin, muscles, subcutaneous tissue and sometimes the viscera, and is present at the time of birth or shortly thereafter. The multiple-lesion type, which is more common, involves either subcutaneous tissue or the viscera together with subcutaneous tissue and the musculoskeletal system. There is a male predominance (1.7:1) in both forms of the disorder[1, 2].

The clinical manifestations and clinical course of IM depend on the number and localization of the tumor lesions[1-3]. A single, large tumor with multiple smaller lesions may be mistaken for a primary malignancy with metastases. Due to prominent tumor vascularity, skin lesions may resemble hemangiomas.

In cases of pulmonary involvement, the lesion may be seen as a solitary mass or diffuse intestinal inflammation in the chest x-ray[1]. Bone lesions are osteolytic and usually solitary, or as seen in our case, multiple bone lesions may also be present[8]. In addition, it may be thought that the right femoral fracture in our case had occurred during delivery. Neurologic symptoms usually develop because of compression of neural structures by the nodules present in the dura mater, soft tissue or bones[3]. Although diarrhea and intestinal obstruction due to involvement of the gastrointestinal tract have been reported[9, 10], in our case, as a consequence of multiple intestinal obstructions, a distinct feature of jejunal perforation was evident.

Evaluation of an infant with IM must include a thorough family history, chest x-ray, skeletal survey, computerized tomographic (CT) scan of the body, echocardiography and tumor biopsies. Contrast studies of gastrointestinal or genitourinary tracts should be performed if relevant signs or symptoms are present. Differential diagnosis should be made with soft tissue sarcoma in soft tissue tumors, and in bony lesions with Langerhans cell histiocytosis.

The histology of IM is quite characteristic and usually shows a distinct zoning phenomenon[1, 3]. The peripheral areas consist of spindle-shaped cells arranged in short bundles and fascicles. These cells demonstrate staining of both myoblastic and fibroblastic character. There is often a considerable amount of

fibroblastic character and frequently a large quantity of collagen within the surrounding matrix. The central portions usually consist of less-differentiated cells arranged around prominent vascular spaces in a hemangiopericytoma-like pattern. In addition, using immunohistochemical technique it has been possible to distinguish IM from the adult form[3].

The prognosis of IM is variable. Solitary or multiple lesions without visceral involvement generally have a benign course and spontaneous regression. However, in a patient with multiple lesions and visceral involvement, the mortality and local recurrence rates are high. Surgical excision should be reserved for cases in which vital structures are affected. Death from IM only results from complications related to visceral lesions in the multiple form of the disorder. Pulmonary involvement seems to carry the most grave prognosis[7].

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