Infantile Myofibromatosis

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We report a neonate presenting with a large labial mass with multiple subcutaneous nodules. A diagnosis of multiple secondary neuroblastoma nodules was made initially. However, biopsy showed the tumor to be infantile myofibromatosis.

Case Report

A two day old female baby presented with a hard nodular mass measuring 4 cm x 3 cm in the right labium majus *(Fig. 1),* along with multiple subcutaneous nodules in the left buttock, left arm, the left supraclavicular and right inframammary regions. The labial mass was bulging into the right lateral wall of the vagina with the vaginal mucosa stretched over it. The subcutaneous nodules were firm and mobile. The lesions in the left supraclavicular and right inframammary regions had telangiectatic areas over it and were also fixed to the overlying skin *(Fig. 2).* There was no abdominal mass or bony lesions on clinical examination. A provisional diagnosis of neuroblastoma with skin nodules was made.

The chest X-ray and abdominal ultrasound examinations were normal. Biopsy of the left inframammary nodule showed a benign spindle cell tumor which was thought to be a fibrous histiocytoma. With this diagnosis, the other lesions were also excised. All the lesions showed fairly well-delineated proliferations of spindleshaped cells. Two patterns were noted. The first pattern seen in the periphery of the lesions showed plump, spindle-shaped cells with elongated vesicular nuclei arranged in whorls and interlacing bundles *(Fig.* 3). These cells superficially resembled smooth muscle cells but histochemical stains showed features of smooth muscle and collagenous differentiation. The second pattern in the central part of the lesion showed smaller, oval cells arranged around vascular spaces in a pericytomatous pattern *(Fig.* 4). The two patterns blended smoothly. There were no histological or cytological features of malignancy.

A skeletal survey revealed lytic lesions with peripheral sclerosis in the upper ends of the left humerus and in both femurs *(Fig. 5).* At 12 weeks of age, the baby was doing well except for two new subcutaneous nodules which had developed in the right supraclavicular region and in the anterior abdominal wall. There was no recurrence in the previously excised sites. There was no evidence of any pulmonary or gastroi-

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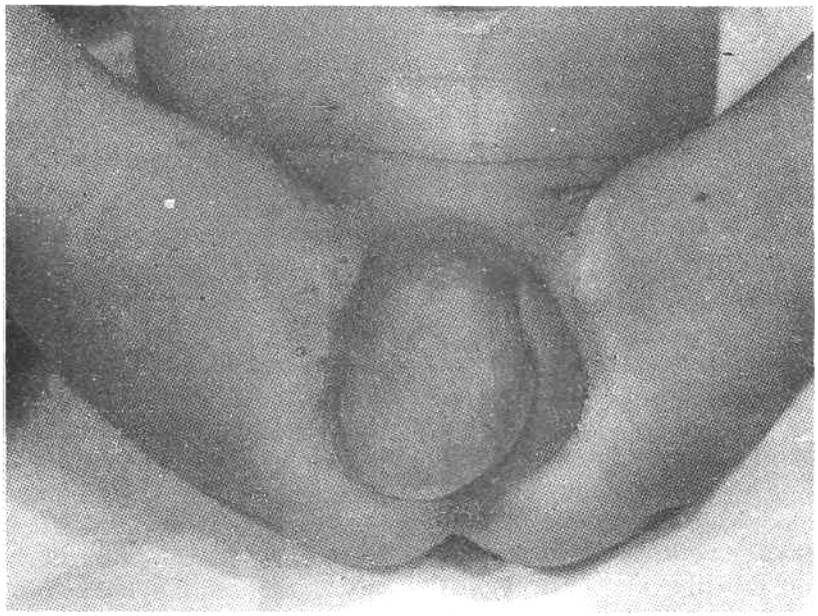

Fig. 1. View of external genitalia showing right labial mass.

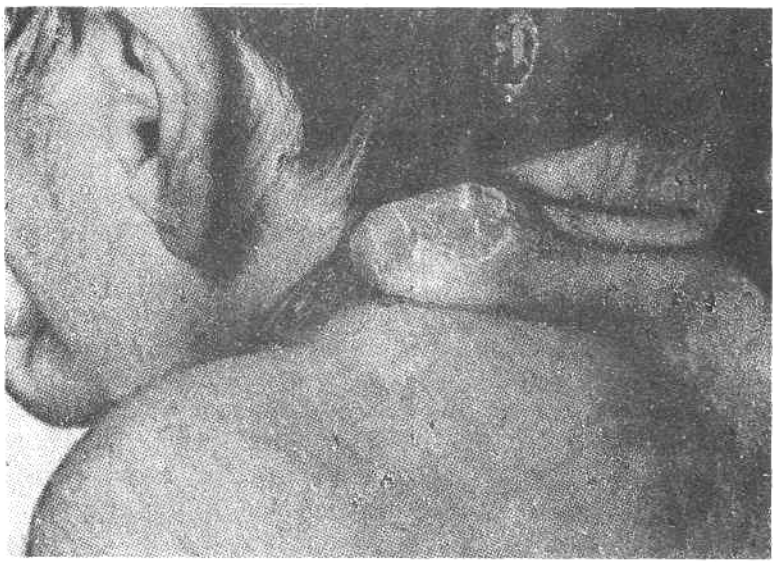

Fig. 2. Nodule in the left supraclavicular region with overlying telangiectasia.

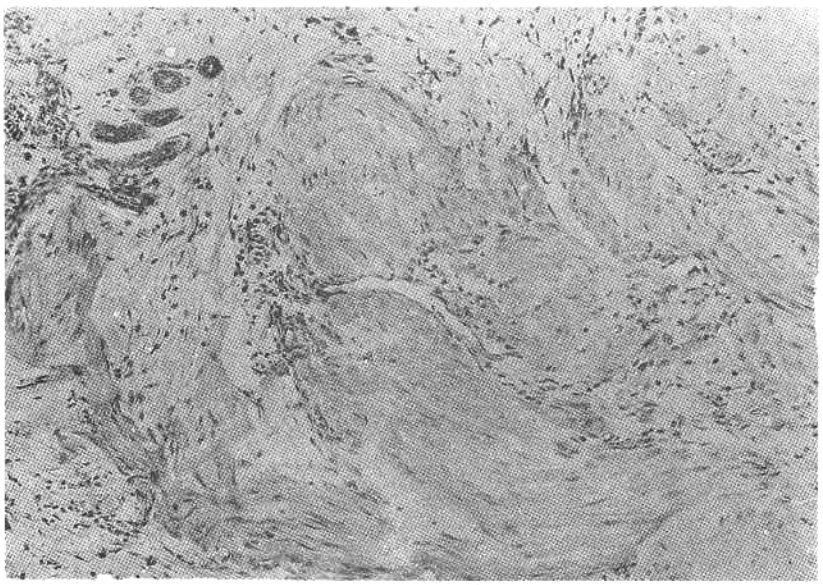

Fig. 3. Cells showing features of myofibroblasts arranged in fascicles and whorls. W & E ×90.

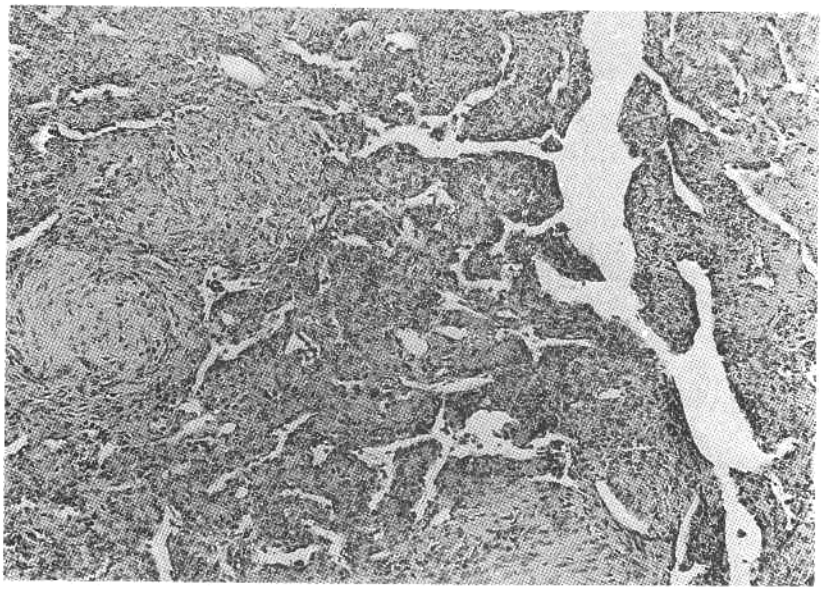

Fig. 4. Hemangiopericytoma–like area showing spindle–shaped cells around vascular spaces. W & E ×40.

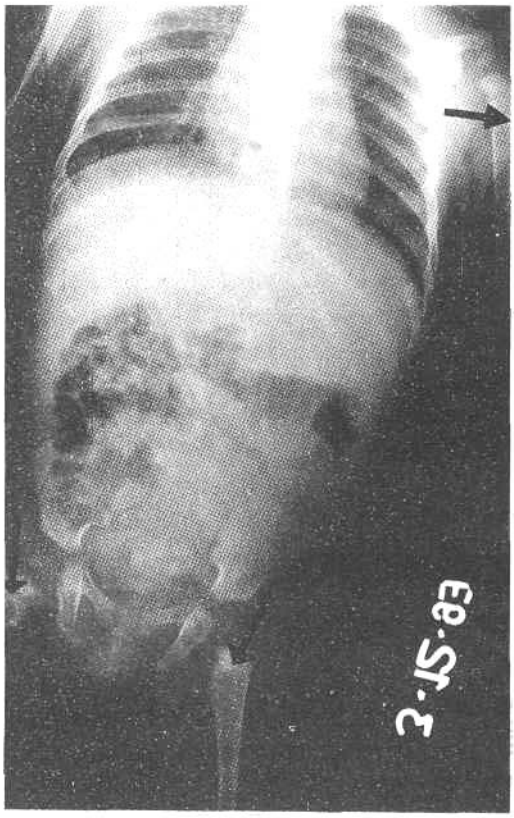

Fig. 5. Skeletal X-ray showing lytic lesions (arrow) in the upper ends of the left humerus and both femurs.

intestinal dysfunction, and the baby is being followed up.

Discussion

Infantile myofibromatosis (IM) was first described by Stout in his article on juvenile fibromatosis in 1954(1). It was described as congenital generalized fibromatosis, multiple mesenchymal hamartomas and multiple vascular leiomyomas of newborn till Chung and Enzinger coined the term infantile myofibromatosis(2).

IM commonly presents in neonates and young- infants although older children and, rarely, adults can also be affected. Classically, the disorder occurs in two forms: a solitary variety occurring in the skin, subcutaneous tissues or muscle; and a multicentric variety involving skin and subcutaneous tissues and bone, with or without visceral involvernent(2,3). The typical presentation is with multiple, well-circumscribed firm, rubbery nodules in the skin, subcutaneous tissue and muscle. One of the nodules may be larger in size than the others, and the picture may resemble that of a primary malignant tumor with multiple metastases(3). Some of the nodules may show a purplish discoloration of the overlying skin, giving the appearance of a hemangioma(2,3). Labial involvement has not been reported previously. Bones commonly affected are the skull bones and metaphyseal regions of the long bones, chiefly the femur and tibia, but the vertebrae, ribs and pelvis may also be affected. They are seen as well-circumscribed lytic lesions with a sclerotic margin, in the roentgenogram(2,4).

When there is visceral involvement, the gastrointestinal tract and the lung are the commonly affected sites, but other viscera like the myocardium, pancreas, omentum, brain, spinal cord, larynx and tongue can also be involved(2,5,6,7). Gastrointestinal lesions, when diffuse, cause a severe watery diarrhea while solitary lesions may produce intestinal obstruction(8,9). Pulmonary involvement gives the appearance of a diffuse interstitial pneumonia or bronchopneumonia on the chest roentgenogram, and causes respiratory distress(2). Spinal lesions can cause cord

compression. IM has been occasionally reported to occur in families and both autosomal dominant and autosomal recessive modes of transmission have been proposed(10,11).

Microscopically, the nodules consist of bundles of plump, spindle-shaped cells in the periphery of the lesion displaying staining characteristics intermediate between fibroblasts and smooth muscle cells arranged in short fascicles resembling a leiomyoma. In the centre of the nodule, necrosis or a hemangiopericytoma-like pattern is seen. Intravascular growth is occasionally encountered(2).

IM must be considered in the differential diagnosis when an infant presents with multiple subcutaneous nodules. When there is no visceral involvement, the disease is benign and self limiting and no active treatment is necessary. Although new nodules may develop, they invariably regress by the age of about two years(4). However, with visceral involvement, the prognosis is worse and death is often due to cardiorespiratory complications or severe diarrhea.

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