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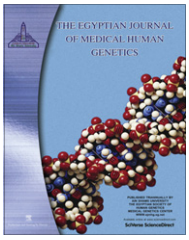

CASE REPORT

Serious life threatening upper airway obstruction in congenital generalized myofibromatosis

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KEYWORDS

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S 100 protein;
APGAR

Abstract We are reporting this patient who was presented with severe upper air way obstruction soon after delivery. She was a girl born with multiple firm nodules, variable in size, widespread all over the body with affection of the oropharynx, long bones, ribs, muscles, subcutaneous tissues, also lungs and intestine. Infantile myofibromatosis was proved on histopathological examination of the masses excised from the oropharynx at the age of 27 days to relieve the upper airway obstruction. The girl had passed the critical stage in NICU and was discharged home with tracheostomy tube. An autosomal recessive mode of inheritance was supported in our case as there was two other male sibs affected with a similar condition and the parents were healthy.

1. Introduction

Infantile myofibromatosis (IM) is described as the most common cause of childhood fibrous tumors; however, its incidence is extremely rare [1]. It was first identified in 1954 by Stout who referred to it as congenital fibromatosis [2]. Several names were used for describing IM including: Multiple mesenchymal hamartoma, multiple vascular leiomyoma of newborn, diffuse congenital fibromatosis, congenital multiple fibromatosis, and multiple congenital mesenchymal tumors. The term IM was first coined by Chung and Enzinger in 1981 [3]. IM may be solitary or multicentric and male to female affection is 1.7/1 [4]. A solitary form describes single nodule in the skin, subcutaneous tissue, muscles or bones, while multiple lesions with or without visceral involvement defines the multicentric form which usually presents at birth [5]. The etiology of the disorder is unknown, it may have sporadic, autosomal recessive, autosomal dominant, or polygenic inheritance and genetic counseling for the family members is important as familial cases were reported as well as chromosomal abnormalities were found in some cases including: del (6)($q^{12}q^{15}$); monosomy 9 q; trisomy 16 q [6,7]. For the surviving babies spontaneous regression is usual (within one to two years) leaving lytic lesions, but recurrence may occur and it was reported in two cases after 8 and 15 years [6], so follow up is important. The prognosis is poor when several internal organs are affected, and it was reported in the literatures that about one third of such infants die in the first 4 months of life from vital organ dysfunction, failure to thrive or infection [8]. Differential diagnosis of soft tissue lesions includes: fibromatosis, desmoid tumors, infantile hemangiopericytoma, fibrosarcoma, and neurofibromatosis; while bonny lesions must be differentiated mainly from Langerhans

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cell histiocytosis, metastatic neuroblastoma, lymphangiomatosis, familial non-osteogenic fibroma, and fibrous dysplasia [9–11]. The characteristic histopathology of, the nodules consists of (1) cells intermediate in appearance between fibroblast-like spindle cells and plump fusiform cells resembling smooth muscle cells, being arranged in short bundles or fascicles and (2) a prominent vascular pattern that is more centrally located and often resembled a hemangiopericytoma. In general, the nodules are well circumscribed, but may be lobulated and have a whorled or nodular appearance. At the periphery, they are composed of curving bundles or intertwining fascicles of spindle shaped or fusiform cells having oval to elongated nuclei, often with rounded or blunt ends, and eosinophilic to clear cytoplasm with fairly well defined cytoplasmic membrane. Delicate wavy bundles of collagen fibers frequently separate the cellular aggregates. In some cases the center of the lesions is irregularly hyalinized, with focally hemorrhagic, or cystic degeneration and coagulation necrosis, often with foci of calcification at its periphery. Occasional deposits of hemosiderin pigment and siderophage are encountered near the necrotic area [3].

Treatment of infantile myofibromatosis includes surgical excision of the lesions interfering with vital organ functions. In cases of recurrence or persistent nodules, treatment trials include: chemotherapy, radiation and steroids which have limited success due to the non-malignant nature of disease [1,12] also interferon alfa was used subcutaneously to treat lesions in a case of Turner syndrome and showed regression of the lesions [13].

2. Case presentation

Our patient was a girl the product of full term (38 weeks gestation), delivered by emergency lower segment caesarian section due to foetal distress. APGAR score was 7 and 8 at the age of 1 and 5 min respectively; birth weight was 2.11 kg, (below the third centile of weight for age). The mother was healthy, para 2 + 1, had uncomplicated pregnancy after non-consanguineous marriage. She had one previous abortion at 2 months and she had two sons one of them had two subcutaneous lesions resolved completely at the age of two years; the other one had cutaneous and visceral lesions (affecting internal structures) which compressed the lumbosacral plexus causing left foot drop. Our case developed severe respiratory distress soon after birth due to upper airway obstruction. Endotracheal tube was inserted with difficulty and the baby was connected to mechanical ventilation and admitted to NICU. There was multiple firm nodules with different sizes, ranging from 0.5 to 3 cm in diameter, in different parts of the body; subcutaneous, fixed to long bones, muscles, ribs, (Fig. 1) and visceral lesions were found in the lungs, intestine and liver. Trials of extubation failed several times due to severe respiratory distress after extubation. Finally we disconnected the baby from the ventilator and endotracheal tube was kept in place to keep patent airway under O2 hood (this maneuver relieved respiratory distress). Surgical excision of the mass in the oropharynx was done and tracheostomy tube was inserted. Initially the baby was kept without oral feeds as there was excessive gastric aspiration, due to partial intestinal obstruction, then feeds were gradually increased to full requirement and despite of high caloric intake the baby was failing to thrive with the weight, height, and head circumference below the 3rd

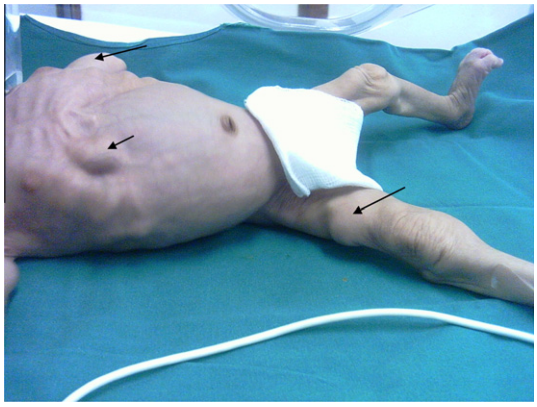

Figure 1 Myofibromatous lesions in the chest and right thigh.

centile for age and her weight was only 2.416 kg at the age of 150 days with gain of only 316 g since birth. Serum electrolytes, kidney function tests and liver function tests were normal. Blood gas analysis showed compensated respiratory acidosis. Skeletal survey showed multiple lesions affecting the long bones of the upper and lower limbs and there was pathological fracture in the upper end of left humerous. MRI showed oropharyngeal masses, multiple nodules in both lungs, and small mass in the liver at the portahepatis. Barium meal and follow through showed delayed passage of the dye due to partial intestinal obstruction. Histopathological examination of the masses excised from the oropharynx (at the age of 27 days) showed: characteristic features of infantile myofibromatosis (Figs. 2 and 3). There was proliferation of thin walled vessels, mitotic count of 14/50 high power fields, entrapment of native ducts, and surface ulceration. The cells were diffusely positive for smooth muscle actin and displayed rare positivity for desmin and were negative for CD34 (surface glycoprotein and functions as a cell–cell adhesion factor), CD 117 (protooncogene c), and S100 protein (specific protein involved in protein phosphorylation).

DIAGNOSIS was infantile (congenital) myofibromatosis.

3. Discussion

We are reporting this rare case of infantile myofibromatosis which was presented with severe upper airway obstruction

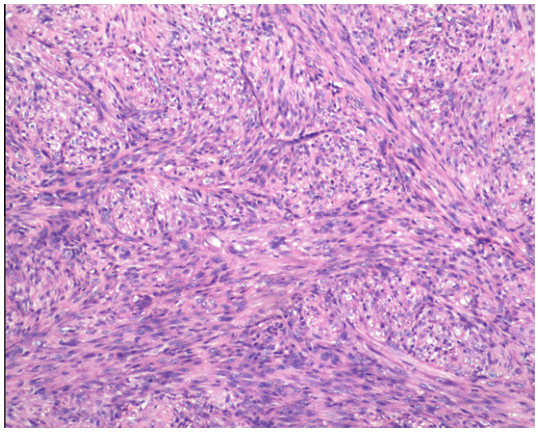

Figure 2 Fascicles of spindle cells (H&E, 40×).

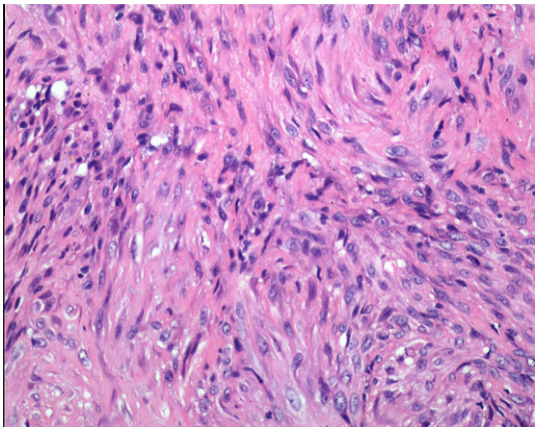

Figure 3 Spindle cells with intervening collagen (H&E, 100×).

soon after delivery, the baby had cutaneous and visceral lesions which represent the multicentric form of the disease [4]. As reported in literatures the lesions can affect the function of vital organs [5]. Our patient was suffering from two important complications which were upper airway obstruction due to oropharyngeal mass (this was relieved by surgical intervention) and partial intestinal obstruction due to another lesion (improved by time with no surgical intervention). There was also pathological fracture in the lower end of the left humerus healed by time. The lesion in portahepatis did not cause obstruction of the common bile duct so there was no surgical intervention. As mentioned in literatures there is chance of spontaneous regression [6] and this occurred in one of the sibs of the baby who had subcutaneous lesions. As the patient had two affected sibs with similar lesions and parents were healthy, the possibility of autosomal recessive pattern of inheritance was raised in our patient. The mortality is high when there are visceral lesions [8]. Our patient bypassed critical situations in NICU and was discharged home at the age of two months with tracheostomy tube having mild respiratory distress, and was on full oral feeds.

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