# Infantile myofibromatosis in a newborn: a case report

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**SUMMARY: Çağlayan F, Çakmak M, Bozdoğan Ö, Abasıyanık F. Infantile myofibromatosis in a newborn: a case report. Turk J Pediatr 2003; 45: 59-63.**

**Infantile myofibromatosis presents as a firm, nodular mass in soft tissues, muscles, or visceras which can be solitary or multicentric, and it may regress spontaneously. We present a one-day-old boy who was admitted to the hospital for two masses, with one below the umbilicus that looked like a hemangiomatous structure and the other in the abdominal skin as a subcutaneous nodule. There was no intraabdominal involvement, and both of the masses were resected at 10 days of life. The one-year follow-up was uneventful.**

***Key words: infantile myofibromatosis, abdominal wall, newborn.***

Infantile myofibromatosis (IM) is a rare condition which generally appears in neonates and infants. Subcutaneous nodules which are the result of myofibroblastic proliferation are mostly located in the head, neck and trunk as a solitary lesion or in multicentric form with or without visceral involvement. Solitary lesions may resolve spontaneously whereas visceral ones have poor prognosis[1]. We present a newborn with IM in the abdominal wall resembling a hemangioma.

## Case Report

A 3700 g male newborn was born at term by vaginal delivery after an uncomplicated pregnancy. There was no consanguinity between the parents. The patient was referred by a private physician whose diagnosis was an abdominal wall defect and hemangioma. Physical examination revealed a suprapubic hemangiomatous mass and subcutaneous nodule in abdominal wall skin near the umbilicus (Fig. 1). A 5x4x3 cm mass was located in the suprapubic region and composed of two pieces. The upper part of the mass had a hemangiomatous appearance. The second mass was a 1.5x1x1 cm subcutaneous nodule near the umbilicus. This was a firm nodule fixed to the skin. Physical examination and vital signs were normal except the finding of the two masses. Complete blood count and biochemical analysis were in normal limits. Roentgenographic skeletal surgey, ultrasonography and computerized tomography (CT) of the abdomen were normal and there was no visceral involvement. Chest X-ray showed no pathology or mass in the pulmonary system. Both of the masses were resected at the tenth day of life. Microscopically, two distinct morphologic features were observed. One revealed plump, spindle-shaped cells which were superficially reminiscent of smooth muscle cells. These cells were arranged in nodules or

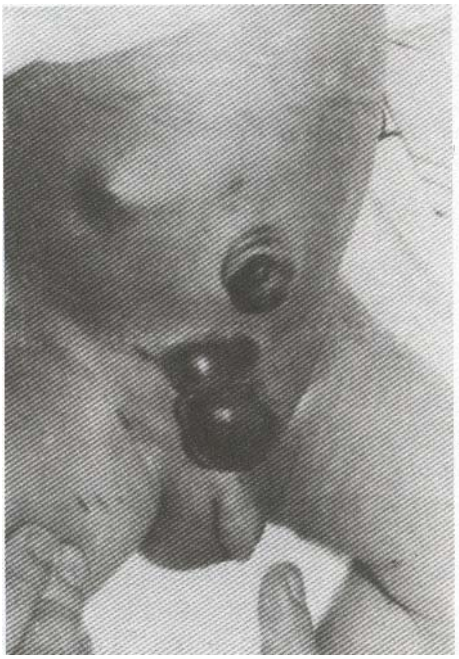

**Fig. 1.** Suprapubic hemangiomatous mass and subcutaneous nodule in abdominal wall.

short bundles (Fig. 2). Around these leiomyoma-like areas, a richly vascular hemangiopericytoma-like pattern was seen (Fig 3). Immunohistochemically, the lesion showed moderate smooth muscle actin (Dako-Clone 1A4) positivity in leiomyomatous areas; diffuse, strong vimentin (Dako-Clone Vim 3B4) positivity was also seen (Figs 4A and 4B, respectively). Based on these morphologic and immunohistochemical findings, the tumor was diagnosed as infantile myofibromatosis. One-year follow-up of the patient was uneventful.

## Discussion

Infantile myofibromatosis was first reported by Stout[2] as congenital generalized fibromatosis in 1954. Several names were used for describing IM, including the following: multiple mesenchymal hamartoma, multiple vascular leiomyoma of the newborn, diffuse congenital fibromatosis, congenital multiple fibromatosis, generalized hamartomatosis, and multiple congenital mesenchymal tumor. The term infantile myofibromatosis was first coined by Chun and Enzinger[3] in 1981.

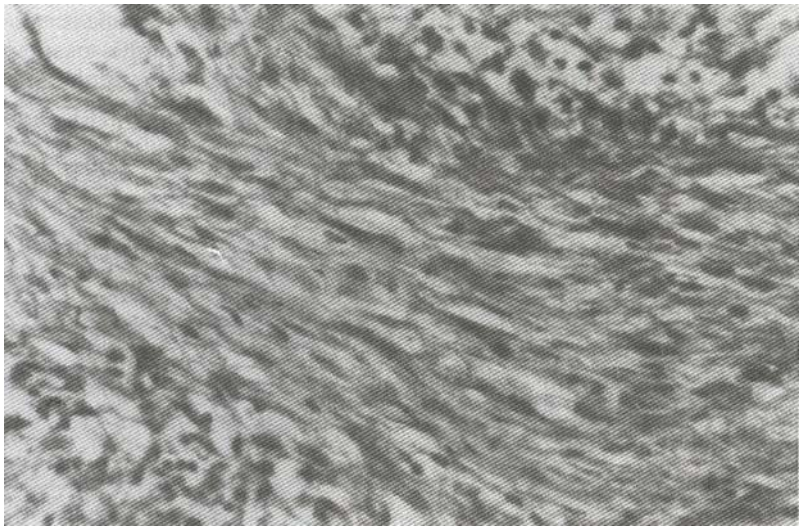

**Fig. 2.** Bundles of well differentiated smooth muscle fibers (myomatous areas) (H.E. X 200).

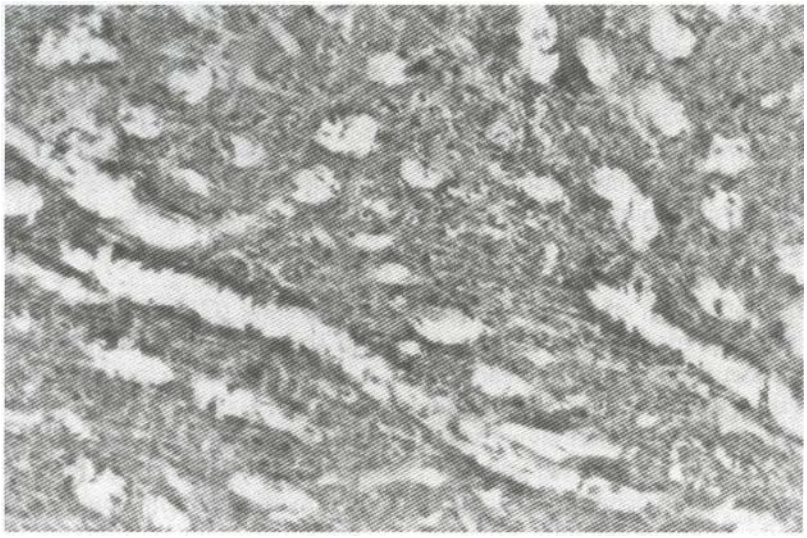

**Fig. 3.** Hemangiomatous areas forming cavernous strustures (H.E. X 100).

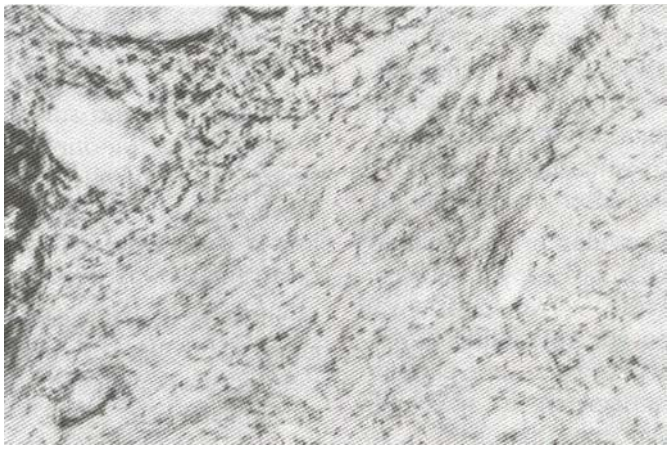

**Fig. 4a.** Moderate smooth muscle actin positivity is present in leiomyomatous area (SMA X 200).

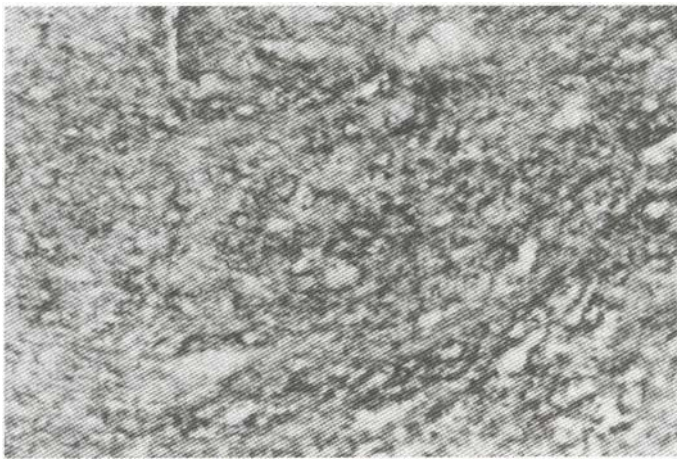

**Fig. 4b.** The cells are diffusely positive for vimentin (Vim X 400).

Infantile myofibromatosis (IM) may be solitary or multicentric and there is a male predominance (1.7/1)[4]. A nodule in the skin, subcutaneous tissue, muscle or bone is defined as the solitary form. Multiple lesions, with or without visceral involvement, defines the multicentric form. Thunnissen et al.[5] reported that the multicentric form usually presents at birth and that the solitary form may present later. Wiswell et al.[4] reported that 37% of multicentric localization had visceral involvement.

Common localizations of the nodules are head and neck, extremities and trunk. Although these locations are usual, some unusual locations such as the pancreas, liver, omentum, and gall bladder have been reported[6-8].

Etiology of IM is uncertain. Autosomal dominant, recessive and polygenic modes of inheritance have been reported[9,10]. Although in utero estrogenic effects have been described in the etiology, this does not explain IM appearing after birth[9]. Hamartomatous origin was also described by Liew et al.[11].

Fibromatosis, desmoid tumors, infantile hemangiopericytoma, fibrosarcoma, and neurofibromatosis must be remembered in the differential diagnosis. Langerhans cell histiocytosis, metastatic neuroblastoma, lymphangiomatosis, familial non-osteogenic fibromata, neurofibromatosis, and fibrous dysplasia should be kept in mind in bone lesions[12-15]. As seen in our case, vascularity of the skin lesions may lead to a confusion in the diagnosis between IM and hemangiomas[4].

Although spontaneous regression occurs in most cases, unless the vital organs are affected, careful follow-up must be done, as recurrences after regressions may occur[4]. Chung et al.[3]

reported a 7% recurrence rate. Recurrences after 8 and 15 years have been reported[9]. Massive apoptosis has been suggested as a mechanism of tumor regression[16]. Spontaneous regression occurs in solitary cases. Our preference for surgical excision was to avoid infection and the possible large defect size after regression.

In cases of persistent nodules or recurrence, steroid, radiation, and chemotherapy have been tried for treatment of IM[1,17]. Two months of subcutaneous interferon alfa (3 million U/m$^2$ daily) treatment in a Turner's syndrome patient with IM resulted in a decrease in size and apoptosis on histological examination[18].

Chung et al.[3] reported 61 cases of IM in 1981. Thirty-seven of them were under five months of age. Five cases of abdominal wall involvement, including four solitary and one multicentric IM located in the abdominal wall, were in this report. As the classification of the ages began with 0-5 months, we do not know whether or not these cases were newborn. Similarly, in Wiswell's report[4] of 170 cases there was no mention of abdominal involvement. However, skin lesions were predominant in most of these cases and so abdominal wall involvement may have been in this category. There was abdominal wall involvement in our case who was newborn.

Twenty-six cases were identified from the literature who were described as newborns in the articles. Fifteen of them were boys and 11 were girls. Bone lesions were prominent in 17 of the cases. Eleven had skin and subcutaneous involvement, including one case in the adductor muscle, two cases of intrapelvic masses, one case of a periorbital mass, one case of the neck, two cases of pulmonary nodules, and one case of a thoracic mass[5,6,19-21]. Major anomalies have been reported, including esophageal atresia, annular pancreas, sacral vertebrae and hypoplastic kidney[22].

Seven newborns had gastrointestinal system involvement and five of them died. Intestinal obstructions and perforations were the main complications in these cases. Newborns with extraabdominal solitary or multicentric nodules must especially be evaluated for intraabdominal involvement, with body imaging by ultrasound, CT, or magnetic resonance imaging (MRI) Ultrasonography and CT of the abdomen were normal and there was no visceral involvement in our case. Another poor prognosis criteria is cardiopulmonary involvement which has a high mortality rate.

Although skin and subcutaneous involvement is common in IM, our literature survey did not reveal abdominal wall involvement in a newborn with similar size and localization as in our case. One abdominal wall involvement in an infant was decribed by Liew et al.[11].

Mentzel et al.[23] described a newborn having congenital clitoral involvement who later developed a 0.6 cm skin lesion in the abdominal and chest wall.

Because of the hemangiomatous appearance, pediatricians and pediatric surgeons must be aware of multicentric localization and the possibility of visceral involvement of IM. Delay in diagnosis and therapy seems important especially in visceral ones as they threat on life in newborns.

**REFERENCES**

1. Burton JL, Lovell CR. Disorders of connective tissue. In: Champion RH, Burton JL, Burns DA, Breathnach SM (eds). Textbook of Dermatology (6th ed) Vol.3. Malden: Blackwell Science; 1998; 2048-2049.
2. Stout AP. Juvenile fibromatosis. Cancer 1954; 7: 953-978.
3. Chung EB, Enzinger FM. Infantile myofibromatosis. Cancer 1981; 48: 1807-1818.
4. Wiswell TE, Davis J, Cunnigham BE, Solenberger R, Thomas PJ. Infantile myofibromatosis. The most common fibrous tumor of infancy. J Pediatr Surg 1988; 23: 315-318.
5. Thunnissen BT, Bax NM, Rövekamp MH, Beek FJ, van Gorp J.Infantile myofibromatosis: an unusual presentation and a review of the literature. Eur J Pediatr Surg 1993; 3: 179-181.
6. Marks MK, Dewan PA, Stokes KB, Smith AL, McKelvie P. Infantile myofibromatosis causing biliary and pancreatic obstruction: a case report. Med Pediatr Oncol 1988; 16: 363-365.
7. Narasimharao KL, D'Cruz AJ, Patel RV, Narasimhan KL, Bannerjee CK, Mitra SK. Infantile myofibromatosis of omentum. Z Kinderchir 1988; 43: 50-51.
8. Goldberg NS, Bauer BS, Kraus H, Crussi FG, Esterly NB. Infantile myofibromatosis: a review of clinicopathology with perspectives on new treatment choices. Pediatr Dermatol 1988; 5: 37-46.
9. Bracko M, Cindro L, Golouh R. Familial occurrence of infantile myofibromatosis. Cancer 1992; 69: 1294-1299.
10. Venencie PY, Bigel P, Desgruelles C, Jacob LS, DufierJL, Saurat JH. Infantile myofibromatosis. Report of two cases in one family. Br J Dermatol 1987; 117: 255-259.
11. Liew S, Haynes M. Localised form of congenital generalized fibromatosis: a report of 3 cases with myofibroblasts. Pathology 1981; 13: 257-266.

12. Aquirre VC, Dominguez R, Calk JB, Gallardo G. Congenital generalized fibromatosis. Ann Ophtalmol 1990; 22: 217-221.
13. Stewart RE, Grosmann DM, Shulkin BL, Shapiro B. Iodien-131 metaiodobenzylguanidine uptake in infantile myofibromatosis. Clin Nucl Med 1989; 14: 344-346.
14. Davies RS, Carty H, Pierro A. Infantile myofibromatosis-a review. Br J Radiol 1994; 67: 619-623.
15. Kotiloglu E, Göğüş S, Ruacan Ş, Akyüz C, Büyükpamukçu M, Sarıalioğlu F. Infantile myofibromatosis: a case with unusual features and review of the literature. Turk J Pediatr 1996; 38: 527-532.
16. Fukasawa Y, Ishikura H, Takada A, et al. Massive apoptosis in infantile myofibromatosis: a putative mechanism of tumor regression. J Pathol 1994; 144: 480-485.
17. Kiel KD, Suit HD. Radiation therapy in the treatment of aggressive fibromatosis (desmoid tumors). Cancer 1984; 54: 2051-2055.
18. Savaşan nS, Fulgenzi LA, Rabah R, Mohamed AN, Ravindranath Y. Generalized infantile myofibromatosis in a patient with Turner's syndrome: a trial of interferon-α. J Pediatrics 1998; 133: 694-696.
19. Wada A, Akiyama HA, Seki HA, Ichihara KA, Miyawaki T, Koizumi S. Spinal canal involvement in infantile myofibromatosis: case report and review of the literature. J Pediatr Hematol Oncol 1998; 20: 353-356.
20. Linder JS, Harris G, Segura AD. Periorbital infantile myofibromatosis. Arch Ophthalmol 1996; 114: 219-222.
21. Bellman B, Wooming G, Landsman L, Penneys N, Schacher LA. Infantile myofibromatosis: a case report. Ped Dermatol 1991; 8: 306-309.
22. Michel M, Ninane J, Claus D, Gosseye S, Wese FX, Moulin D. Major malformations in a case of infantile myofibromatosis. Eur J Pediatr 1990; 149: 251-252.
23. Mentzel T, Calonje E, Nascimento AG, Fletcher CD. Infantile hemangiopericytoma versus infantile myofibromatosis. Study of a series suggesting a continuous pecturm of infantile myofibroblastic lesions. Am J Surg Pathol 1994; 18: 922-930.