Case Report

Juvenile Fibromatosis: Report of a Rare Case

Halima Oziohu Aliyu, Tunde Talib Sholadoye[1], Saidu Umar Abdulkarim[2]

Department of Pathology, Ahmadu Bello University, Ahmadu Bello University Teaching Hospital, [1]Division of Paediatric Surgery, Department of Surgery, Ahmadu Bello University, Ahmadu Bello University Teaching Hospital, [2]Department of Surgery, Ahmadu Bello University Teaching Hospital, Shika, Zaria, Kaduna, Nigeria

Abstract

Fibromatosis is a rare benign tumor arising from the myofibroblast. It is a locally aggressive tumor with a high rate of recurrence but no malignant potential. Fibromatosis can occur anywhere in the body. It is rare in the head-and-neck region and even rarer in children. We report a case occurring in the neck of a child. A 7-year-old boy presented with progressive left-sided, posterior neck swelling of 4 years and 3 years history of the loss of ability to vocalize. Imaging studies revealed a mass at the base of the skull and posterior neck on the left, with the preserved surrounding bone. The patient had a complete excision of the mass and histology confirmed juvenile fibromatosis. Juvenile fibromatosis of the head-and-neck region is a rare benign tumor of childhood. Tissue biopsy confirms the diagnosis. The recurrence rate is high following wide local excision.

Keywords: Desmoid tumor, fibromatosis, head and neck, juvenile

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Introduction

Fibromatosis is a benign but frequently recurrent tumor arising from the connective tissue of muscle, overlying fascia, aponeurosis, or periosteum.[1] It is rare with an annual incidence of 2–4 cases per million, representing 0.03% of all neoplasms.[2] It occurs anywhere in the body but is uncommon in the head-and-neck region and more so in children.[3] The classification of fibromatosis varies deriving their names from their site of occurrence (abdominal and plantar.).[4] However principally, fibromatoses are classified into juvenile and adult fibromatosis which show differences in their site of occurrence and sex predilection.[5] Histopathologic diagnosis and wide local excision with free resection margins are its preferred management.[5] This, however, may be difficult to achieve in the neck with the presence of vital structures and more challenging in children.[6] This report is of a 7-year-old boy with a posterior neck Juvenile fibromatosis tumor.

Case Report

A 7-year-old boy presented with 4-year and 3-year history of a progressive posterior neck mass and poor vocalization. Examination revealed a stable child with a huge firm, nontender mass found in the left posterior-lateral aspect of the neck, extending from below the occiput to just above the scapular vertically; and from the cervical spinous processes to the posterior border of the left sternocleidomastoid muscle, and attached to the underlying structures, but not to the skin [Figure 1a]. His head rotation to the left was limited to 15° from the midline.

Ultrasonography revealed a solid mass with internal echoes suggesting a soft-tissue mass. The radiograph of the neck revealed no bony involvement. Head-and-neck computed tomography scan showed a soft-tissue isodense nonenhancing mass (Hu-72) arising from the base of the skull and posterior neck. The underlying bones were preserved as well as the airway. No intracranial extension was seen. Hematological and other biochemical parameters were within the normal limits. Fine-needle aspiration showed a few foam cells, fibrous stromal fragments, adipocytes, and hemorrhage only. No other masses were present.

Address for correspondence: Dr. Halima Oziohu Aliyu, Department of Pathology, Ahmadu Bello University and Ahmadu Bello University Teaching Hospital, Zaria, Kaduna State.
E-mail: haliyuo77@yahoo.com

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Intra-operative findings were a well-vascularized firm left-sided neck mass extending from posterior border of the left sternocleidomastoid muscle to the posterior neck transversely, and from the posterior nuchal line to the left trapezius muscle (22 cm × 18 cm); firmly attached to the prevertebral fascia at the level of C2/C3 vertebra and weighing 280 g [Figure 2]. The carotid sheath and its contents were free. Gross complete tumor excision was achieved. The postoperative period was unremarkable and the wound healed within 10 days with up to 90° left-sided head rotation from the midline achieved. He was discharged and referred for speech therapy. He has had seven follow-up clinic visits in 3 years with no regrowth [Figure 1b].

Pathological examination revealed a solid gray-white tumor; sections of which showed a focally infiltrative, vaguely nodular, and cellular fibroblastic tumor in a collagenous background. The cells had bland nuclei with no mitotic activity noted [Figure 3]. The tumor focally infiltrated skeletal muscle and the resection margins were free. Stains for desmin and vimentin were positive [Figure 4]. A diagnosis of juvenile fibromatosis was made.

Discussion

Juvenile fibromatosis was first described by Stout as a distinct intermediate lesion arising from the musculoaponeurotic system decades ago.[7] Although more cases have been reported, its rarity has been a limiting factor in its study, especially in children.[6] Peña *et al*. reported that as many as 30% of cases occur in children ranging in age from birth to 16 years.[8] A case report search gave an age range of between 20 months and 6 years,[4,9-12] whereas Miyashita *et al*. gave a broad age range of <19 years.[13] Our patients' tumor was first noticed at the age of 3 years with slow, but progressive growth warranting his presentation at the age of 7 years as it had interfered with his speech and limited his head movement.

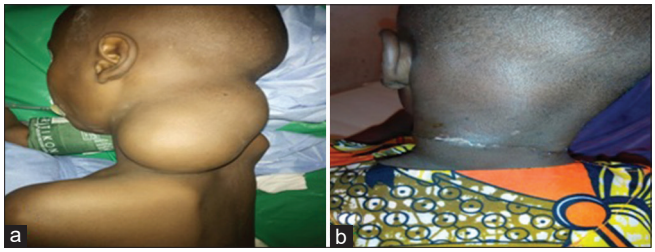

Figure 1: Preexcision view (a) and three weeks post excision view at follow-up clinic visit (b)

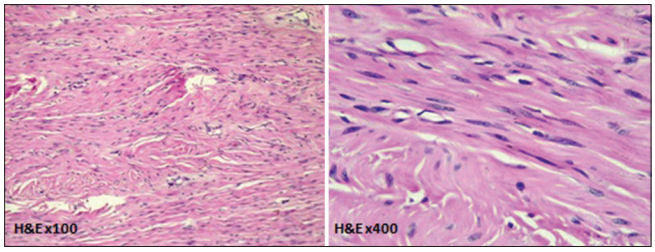

Figure 3: Plump fibroblasts in a collagenous background

Most fibromatoses occur in the abdomen.[6] It has been reported that <2% of all fibromatosis occur in the head–and-neck region and this value further drops when counted in children.[3,4] The most common site of involvement in the pediatric age group is the scalp then the neck usually presenting as a nodular mass in the subcutis as was reported by Kumar *et al*.[4] Our patient presented with a progressive firm neck mass, attached to the underlying structures and free from the skin; consistent with the findings in the literature.[4,5,8]

The cause of fibromatosis is yet elusive; however, certain factors such as genetics (familial adenomatous polyposis syndrome (FAP), and Gardner's syndrome), hormones (regression with anti-estrogen therapy), trauma, and irradiation have been linked to its etiology.[4,6,11,12] These were not demonstrable in this index case.

Other similar tumors apart from presenting in children, differ from juvenile fibromatosis by certain features which include the autosomal recessive, extremely rare, and typically fatal Juvenile hyaline fibromatosis with its tumor-like multiple dermal masses on the head and in soft tissues and bone, whereas juvenile fibromatosis is often solitary and sporadic;[14] and its close resemblance in age, site of occurrence, and presentation to infantile fibrosarcoma that is usually differentiated by some histologic parameters discussed below.[15]

As stated earlier, juvenile fibromatosis closely resembles a malignant tumor. This is due to its propensity to invade adjacent structures but without distant spread;[3] this was evident in

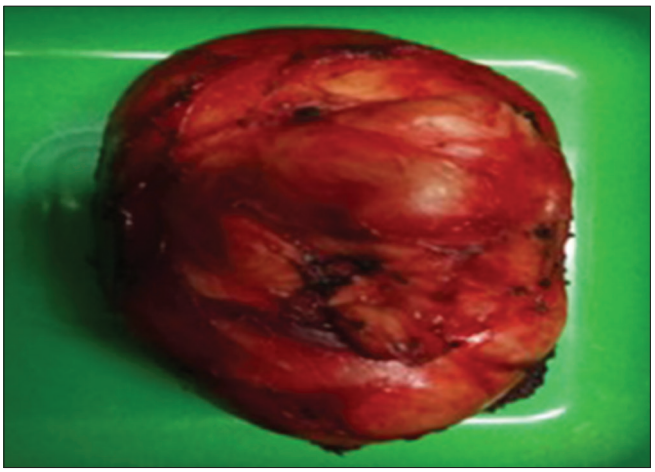

Figure 2: Gross appearance of the excised neck mass

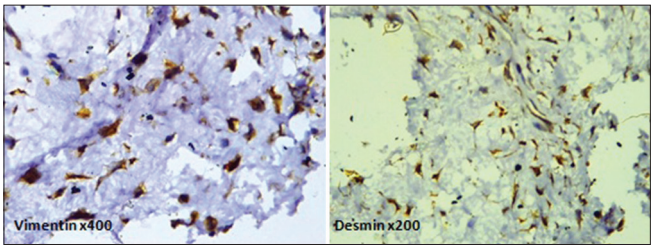

Figure 4: Positivity to vimentin and desmin antibodies

its imaging in the index case. Magnetic resonance imaging and computerized tomography (CT) scan are important in identifying the local extent of the tumor, and more importantly demonstrating the involvement of vital structures such as the carotid sheet; this helps in planning for complete resection with good margins.[9] CT scan of the mass, in this case, was useful in delineating the tumor extent showing no extension to the carotid sheath or bone which enabled its excision. However, it suggested soft tissue and nerve involvement evidenced by poor vocalization from the recurrent laryngeal nerve infiltration. MRI which would have given a better soft tissue delineation of the tumour was not done due to financial constraints. De Riu *et al*. reported a case involving the mandible necessitating a mandibular excision and a long-term follow-up with repeated reconstructions in the growing child;[9] these points out the difficulty usually encountered in the management of this tumor in children.

The pathological examination usually shows a firm gross appearance and a microscopic composition of long fascicles of spindle-shaped cells of myofibroblastic origin that may have background collagen. Mitosis, nuclear atypia, and necrosis are typically not seen, as was in this case. The index tumor showed microscopic infiltration into the adjacent tissues particularly skeletal muscle fibers with resulting muscle atrophy and had mild lymphoid infiltrates. These features are quite different from the numerous hyaline material interspersed between the fibroblasts as is seen in juvenile hyaline fibromatosis,[14] and the focally necrotic, mitotically active hypercellular spindle cell appearance of infantile fibrosarcoma.[15]

Approximately 70%–75% of juvenile fibromatosis show nuclear beta-catenin positivity, especially when occurring in association with FAP.[16] They are usually positive for vimentin and may be positive for desmin, muscle-specific actin (MSA), and smooth muscle actin (SMA),[8,10] both vimentin and desmin were positive in this case. Beta-catenin was not tested for due to its nonavailability at this center, while both MSA and SMA were negative.

Several adjuvant modalities for fibromatosis have been tried and include chemotherapy, radiotherapy,[7] antiestrogen therapy, steroids, and castration; but surgical excision with free resection margin is the treatment of choice.[7] However, this could be challenging in children with short neck and where there is invasion of vital structures. A balance between the adequate excision and preservation of these structures has to be maintained.

Where surgery alone fails, the tumor recurrence tends to be especially infiltrative of the surrounding structures making re-excision destructive and problematic.[13] Adjuvant modalities may then be tried with some successes on record.[13] Miyashita *et al*. reported a recurrence rate of 27.2% following surgery. Seven out of the eight children with tumor at the cervical paraspinal area had a recurrence.[13] The high rate of recurrence seen in the head-and-neck region is attributable to the difficulty in achieving a free resection margin. Radiotherapy was a beneficial adjuvant in them. Thus, patients with R2 resection (resection with gross positive margin/incomplete resection), and R1 resection (resection with positive microscopic margins) or inoperable tumors may benefit from radiotherapy.[4] Follow-up clinic visits for 3 years in this case has shown no recurrence.

Conclusion

Juvenile fibromatosis of the head-and-neck region is a rare occurrence. A high suspicion enables early diagnosis, and a combination of surgery with adjuvant therapies will aid in treating affected children. Local excision with free margins is associated with good results but because of its known infiltrative property, timely follow-up after surgical resection is important in its management.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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