CASE REPORT

Aggressive fibromatosis (desmoid tumour) of the head and neck: a benign neoplasm with high recurrence

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SUMMARY

A 50-year-old man presented with a 5-month history of swelling over the right side of neck. The swelling was associated with dull aching pain radiating to the forearm without associated weakness of upper extremity or sensory loss. There was no history of trauma. On examination a fixed mass approximately 8×6 cm in size, smooth, firm in consistency, with ill-defined margins was present in the right posterior triangle. MRI scan of the neck revealed well-defined, lobulated, heterogeneously enhancing altered signal intensity mass at the root of neck. Debulking of the tumour was performed in view of its close proximity to the brachial plexus. Histopathology revealed aggressive fibromatosis (AF). AF is a benign fibrous neoplasm arising from fascia, periosteum and musculoaponeurotic structures of the body. AF in the head and neck region tends to be locally aggressive with a nature to invade bone and soft tissue structures.

BACKGROUND

Aggressive fibromatosis (AF) is a benign fibrous neoplasm arising from fascia, periosteum and musculoaponeurotic structures of the body. AF in the head and neck region tends to be locally aggressive, and its nature to invade and erode bone and soft tissue makes excision difficult. The presence of vital neurovascular structures in the head and neck makes it even more complicated. It is wise not to compromise on the vital structures considering the benign nature of the disease.

CASE PRESENTATION

A 50-years-old man presented with a 5-month history of a swelling over the right side of neck. The swelling was gradual in onset and progressive in nature. For the past 1 month he had dull aching pain radiating to the forearm. There was no history of trauma. There was no associated weakness of upper extremity or sensory loss. On examination a fixed mass approximately 8×6 cm in size was present in the right posterior triangle, not associated with any overlying skin changes. The mass was found extending superiorly 4 cm below the mastoid tip, medially involving the lateral half of right clavicle and laterally extending to anterior border of trapezius. On palpation surface of the swelling was smooth, firm in consistency, with ill-defined margins, non-tender and non-mobile. Oral cavity examination was normal.

INVESTIGATIONS

Chest X-ray, indirect laryngoscopy was normal.

Fine needle aspiration cytology revealed no malignant cells.

MRI of the neck showed a well-defined, lobulated, heterogeneously enhancing altered signal intensity (T1 isointense, T2 hyper-intense) mass at the root of neck having epicentre in right posterior triangle with multiple areas of calcification and haemorrhage. It extends craniocaudal from C5-D3 level. Anteriorly it was abutting the scalenus medius muscle. Posteromedially it was abutting the paraspinal muscles, and posteriorly abutting the serratus anterior muscle. Anteroinferiorly, it was abutting the posterior surface of posterior aspect of first and second ribs. The lesion was seen to abut the C5, C6 roots and the upper trunk of brachial plexus (figure 1).

Histopathology: The lesion composed of sheets and long sweeping, intersecting fascicles of spindle cells with elongated vesicular nuclei, indistinct nucleoli, long eosinophilic cytoplasmic processes, increased mitotic figures and foci of collagen deposition, proliferating capillaries and mast cell infiltrate suggestive of aggressive fibromatosis (figure 2).

DIFFERENTIAL DIAGNOSIS

- Metastasis of unknown origin
- Neurofibroma

TREATMENT

The patient was planned for wide local excision. A horizontal incision was made over the tumour

Figure 1 MRI of the neck: a well-defined, lobulated, heterogeneously enhancing altered signal intensity (T1 isointense, T2 hyper-intense) mass at the root of neck having epicentre in right posterior triangle with multiple areas of calcification and haemorrhage.

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Figure 2 Histopathology: lesion composed of sheets and long sweeping, intersecting fascicles of spindle cells with elongated vesicular nuclei, indistinct nucleoli, long eosinophilic cytoplasmic processes, increased mitotic figures and foci of collagen deposition, proliferating capillaries and mast cell infiltrate suggestive of aggressive fibromatosis.

and the overlying muscles separated. A cardiothoracic and vascular surgery (CTVS) surgeon was called for help during the surgery. It was not possible to excise the tumour because of its close proximity with the brachial plexus (figure 3). Hence debulking of the tumour was performed, leaving behind a thin rim of tissue adjacent to the nerve roots.

OUTCOME AND FOLLOW-UP

The postoperative period was uneventful and the patient recovered completely. Power of the right hand muscles was 5/5 and reflexes were normal. He was started on tab indomethacin 75 mg once daily, an extended release medication. He is on regular follow-up for the last 3 months. He has been advised to take adjuvant radiotherapy in view of the residual disease. The patient is yet to decide on the same.

DISCUSSION

AF is a benign fibrous neoplasm arising from fascia, periosteum and musculoaponeurotic structures of the body.¹ They are widespread, infiltrative and well-differentiated firm overgrowths of fibrous tissue, and tend to be locally aggressive. They may arise from any fibrous tissue in the body and may present in a variety of different locations. They are most commonly seen in the

Figure 3 Operative specimen: debulking of the tumour.

abdomen (roughly two-thirds of all aggressive fibromatosis). Extra-abdominal aggressive fibromatosis makes up the remaining one-third, of which 11–15% may be seen in the region of the head and neck. Nasal cavity, nasopharynx and paranasal sinus involvement is rare.² AF can be associated with polyposis coli, colonic adenocarcinoma, epidermoid cysts and osteomas.¹

Aggressive variants of fibromatosis seen in the head and neck region (defined as that portion of the body above the clavicle anteriorly, superior to first thoracic vertebral spine posteriorly and above the shoulder girdles laterally) tend to be locally aggressive, invading and eroding bone and soft tissue, thus compromising vital structures.

Incidences of trauma (surgical or non-surgical) have been observed to precede development of fibromatosis in 19–49% cases. The theorised pathogenesis involves abnormal response to healing with persistent immature fibroblast formation which later gives rise to the tumour. Steroid sex hormones are also believed to play a role in pathogenesis.³

Incidence of AF of head and neck appears to be roughly equal in both sexes, with a predilection towards affecting younger patients, especially in the teenage group. Recurrence is often associated with patients in this younger age group, but this is controversial.⁴

On histopathology, the gross appearance of the tumour is that of interlacing trabeculae of mature fibrous tissue with ability to infiltrate muscle, adipose and other fibrous tissue. It tends to encase neurovascular structures with deeper invasion. Histologically it is composed of mature fibroblasts with a collagenous component.⁵ AF is a histopathological diagnosis.

Clinical features of AF are usually the result of pressure effects of the growth on nerves and vascular structures. In cases involving the shoulder region, the most common clinical features include tingling, numbness, paraesthesia and weakness of muscles of the hand (due to compression upon the brachial plexus). ‘Shooting’ pains radiating from shoulder to hand have been reported as well.² Malignant transformation has been known to happen, but is quite rare. Metastasis has not been known to occur.⁶

Imaging is of importance in the diagnosis. On CT it appears hyperattenuating, with significant enhancement on application of contrast material. MRI shows a multilobulated lesion of heterogeneous signal intensity.

Complete surgical excision of aggressive fibromatosis is considered the only effective treatment. However, there is a high chance of recurrence (20–77% in extra-abdominal cases), especially in case of head and neck fibromatosis (40–70%). Majority of recurrences occur within 18 months of excision.⁴

Chemotherapy may have a significant role, considering the possible hormonal etiopathogenesis of the condition. One of the most commonly tried chemotherapeutic drugs is the anti-estrogenic drug Tamoxifen. Other drugs that have been tried for similar effects include toremifene, progesterone, medroxyprogesterone acetate, prednisolone, testolactone, testosterone, desoxycorticosterone and goserelin.^{7 8}

Non-steroidal anti-inflammatory drugs (NSAIDs) may also have a role in treatment. Indomethacin, Sulindac and Colchicine have been tested in cases of AF to varied degrees of success.^{9 10}

In the event that antioestrogen therapy and NSAIDs fail to act sufficiently, cytotoxic drugs such as doxorubicin-dacarbazine combination; doxorubicin-cyclophosphamide-vincristine; or vincristine, actinomycin-D, cyclophosphamide (VAC) regimen may be used. In paediatric patients and in women of childbearing age, methotrexate may be administered in tandem with vinblastine as a less toxic alternative to doxorubicin or VAC regimen.¹¹

Recent advances in treatment such as interferon (IFN) α and tyrosine kinase inhibitor Imatinib have come to the fore. *In vitro* studies have shown that there may be an antiproliferative effect by IFN α and retinoic acid on fibroblasts, while IFN γ may show inhibitory effects on fibroblast collagen synthesis. IFNs may be considered for adjuvant therapy in cases of surgical excision with a view to prevent recurrence. There may, however, be IFN-related side effects.¹² Tumour expression of activated receptor tyrosine kinase c-kit and platelet-derived growth factor receptor α provides scope for another therapeutic option.¹³ Irradiation has little effect barring transient reduction in size of tumour in some cases. As recurrences may occur up to 12 years after initial treatment, lifelong follow-up is a necessity.

Learning points

- ▶ Aggressive fibromatosis (AF) is a benign fibrous neoplasm arising from fascia, periosteum and musculoaponeurotic structures of the body.
- ▶ AF of the head and neck region tend to be locally aggressive, invading and eroding bone and soft tissue, thus compromising vital structures.
- ▶ Complete surgical excision is considered the only effective treatment.
- ▶ High chance of recurrence in case of head and neck fibromatosis (40–70%).
- ▶ Adjuvant treatment options include Tamoxifen, non-steroidal anti-inflammatory drugs, cytotoxic drugs, interferon and Imatinib.

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Competing interests None.

Patient consent Obtained.

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