

CASE
REPORT

Desmoid Fibromatosis of Submandibular Region

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INTRODUCTION

Desmoid tumors are benign musculoaponeurotic tumor accounting for almost 0.03% of all neoplasms. Despite their benign nature, they are known as aggressive fibromatosis, with the potential to cause damage to surrounding structures, leading to organ damage. It is caused by mutations of fibroblast cells that are found throughout the body. Fibroblasts play crucial role in wound healing and protection of vital organs such as lungs, liver, blood vessels, heart, and kidneys. When mutations, either sporadic or inherited occur in these cells, they become neoplastic and lead to formation of desmoid tumors. Thus, aggressive fibromatosis is a monoclonal proliferative disease with inability to metastasise.^[1]

Despite their purely benign histological appearance and insignificant potential to metastasise; the tendency of desmoid to cause infiltration is of a great concern in the context of deformity, morbidity and mortality resulting from pressure effect on cellular level up to the obstruction of vital structures and organs. Besides that, the high prevalence of recurrence makes the treatment of these rare fibrous tumors challenging. Moreover, the depth of invasion of the tumor is associated with the rate of recurrence.

ABSTRACT

Desmoid fibromatosis is a benign yet locally aggressive tumor with a tendency to recur. It causes considerable morbidity particularly when it arises in a small area in the head and neck region. This tumor is extremely rare in the submandibular region. We report a case of desmoid tumor in the submandibular region in a 32-year-old male who presented with right submandibular swelling postextraction of right lower wisdom tooth. Excision biopsy was carried out initially following inconclusive fine needle aspiration and discussion at multidisciplinary meeting. The tumor recurred 4 months following initial excisional biopsy necessitating a more radical secondary approach involving segmental mandibulectomy. Intraoperatively we also noted that the tumor was originating from the site of previous wisdom tooth extraction, raising the question of surgical trauma as precursor of desmoid tumor. We achieved a negative resection margin and a complete remission for 24 months.

Key words: Desmoid fibromatosis, mandibulectomy, submandibular region

There is no standardized classification for desmoid tumor, but the terms, abdominal and extra-abdominal fibromatosis by World Health Organization (WHO) are usually employed in a wider context.^[2] Adult extra-abdominal fibromatosis can arise in any anatomic site, commonest being the limbs, shoulder, thigh, and buttock. Incidences in the head and neck are uncommon and accounts for 5-10% of all soft tissue tumors.^[3] Extra-abdominal tumors occurs more sporadically and often can be treated effectively by local resection with systemic treatment reserved for refractory tumors.^[4] They are frequently aggressive, with poorly circumscribed local infiltrative pattern and tendency to recur.

In contrast, systemic therapy is usually considered as first line treatment of intraabdominal desmoids, because they are often diffusely infiltrative and surgically unresectable.^[4]

Previous studies on desmoid tumors have mostly dealt with the intraabdominal and thoracic regions but very few actually discussed the repercussions and management of such benign tumors arising in small regions such as head and neck. With tumors in this region, patients usually present with trismus, ulceration, intraoral bleeding, otalgia, dyspnea, dysphagia, and proptosis but its most important

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pathogenic property is the invasion of the compact neurovascular and anatomical structures.^[5]

There have been few reported cases of desmoid fibromatosis in the submandibular region, but to date no consensus has been reached as to the optimal treatment of this tumor in this small region. So, the primary focus of this case report study is to discuss our case of desmoid fibromatosis in the submandibular region, treatment options and literature review of recurrent desmoid tumor in the submandibular region.

CASE REPORT

An otherwise healthy 32-year-old male presented to our institution with 6 weeks history of right submandibular swelling postright wisdom tooth extraction. There was no fluctuation of the size of swelling and no postprandial changes. The swelling was increasing in size gradually. He had no other associated symptoms.

Head and neck examination revealed a firm 4 × 3 cm mass in the right submandibular region. There was no associated lymphadenopathy and the rest of the examination was normal.

Ultrasound examination was requested and fine needle aspiration (FNA) of the mass was carried out. Ultrasound examination revealed a 3.5 cm ill-defined mass of uncertain origin and FNA was inconclusive as there was uncertainty about the nature of the swelling, whether it was benign or malignant.

Following multidisciplinary meeting, excision biopsy of the mass was carried out. Histology of the postoperative specimen was complex. Questions were raised whether it was a Chronic Sclerosing Sialadenitis, Kuttner tumor or Desmoid Fibromatosis.

Histopathological slides were reviewed again in the multidisciplinary meeting. Specimen showed spindle lesions within muscles with abundance inflammatory cell and classic myofibroblastic cells [Figure 1] and final diagnosis of desmoid fibromatosis was made.

Patient was followed up regularly on 1-monthly basis. Four months postoperatively, the tumor recurred in the same place. This time, patient underwent magnetic resonance imaging (MRI) scanning of the head and neck region and also panorex view of the mandible. MRI confirmed infiltrative mass in the right submandibular region pushing the tongue off midline. There was no evidence of

muscular invasion [Figure 2]. The panorex view showed an indentation on the right side of the angle of the mandible, which at that point of time was thought to represent focal impression by the submandibular mass [Figure 3].

Following further discussion in the multidisciplinary meeting, patient was booked in for reexcision of the mass. This time access to the neck was gained through modified Blair incision. Tumor was noted to involve the outer cortex of the body of the mandible [Figure 4] and interestingly, originating from the socket of wisdom tooth (tooth was removed five months ago). Segmental mandibulectomy was carried out sparing the inferior alveolar nerve [Figure 4]. Tumor was resected out together with the involved portion of the mandible. Inner cortex of the tooth socket was also removed. Drain (3/4 inch) was inserted and wound was closed in layers. Functions of facial, lingual and marginal mandibular nerves were all preserved and patient had normal jaw movement. He was discharged day 3 postsurgery. Histology of resected specimen was confirmed to be desmoid fibromatosis with clear resection margin. He was seen 2 and 6 weeks postsurgery and was last seen 24 months ago and remained disease free.

DISCUSSION

Although considered to be benign, the fibroblastic proliferation of desmoid tumors is indefinite and the tumor itself has an unpredictable clinical course. Microscopically, the tumor arises from myofibroblasts, lacks true capsule and usually infiltrates into surrounding normal structures.^[6] Desmoid tumors can develop in any site of the body and pathologically appears to be identical. Superficial desmoids are less aggressive and may present as slightly painful lump. The deep ones are more aggressive and have the tendency to press and rupture critical structures such as nerves and blood vessels. These effects are critically exacerbated when they arise in smaller regions such as head and neck.

Our patient presented with painless lump in the submandibular region, 6 weeks postextraction of right wisdom tooth. The tumor recurs approximately 4 months after initial resection necessitating a more radical secondary approach. Surgical trauma has been implicated as the precursor of desmoid tumors in approximately 68-86% of abdominal and extra-abdominal desmoids.^[7,8] We suspect extraction of the wisdom tooth could have been the precursor that have propagated into the initial mass and incomplete clearance around the mandible and the tooth socket could have been the cause of recurrence, but this is still to be proven.

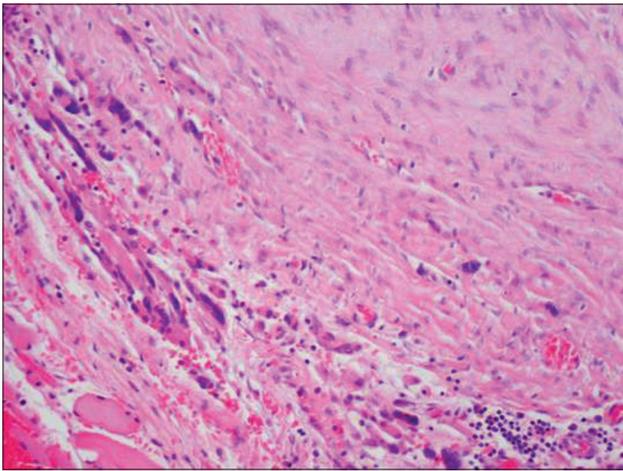


Figure 1: Medium power: Spindle cells with classic myofibroblastic cells within collagenous to myxoid stroma and associated inflammatory cells. No atypical mitosis or anaplastic elements are seen. (Note: Proliferation of spindle cells with bland nuclei and fascicular arrangement seen)

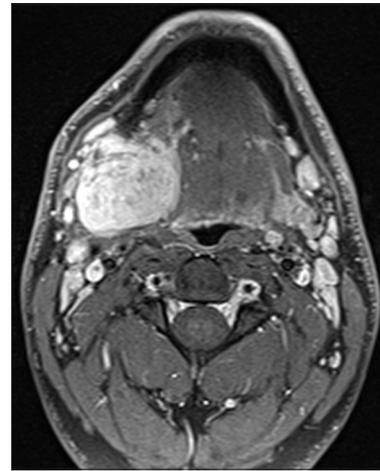


Figure 2: Axial cut of MRI scan showing large mass in the right submandibular region, indenting and pushing the tongue off midline



Figure 3: Panorex view of the mandible



Figure 4: Intraoperative photograph showing tumor attachment to the mandible. Segmental mandibulectomy was carried out sparing the inferior alveolar nerve

The local control of this tumor constitutes continuing treatment dilemma and the clinical management remains a challenge. Even following complete resection of the tumor with clear margin, local failure is common. Reported recurrence rates vary from 25% in cases of complete excision in patients with head and neck desmoid tumors to 80% in patients who had incomplete excision.^[9]

Surgery with wide margin has conventionally been the therapeutic pillar for primary localized desmoid tumor. The recommended margins are 2 and 4 cm, respectively, in the transverse plane and along the longitudinal axis and less than 3 cm for small tumors.^[6] This recommended margin may not be easily applicable in a compact region of head and neck and can be mutilating and results in functional and cosmetic loss. Surgical has to be tailored to what is attainable in terms of margin while preserving the vital structures and their function.

As the goal of treatment is for complete excision and minimal recurrence, Desmoid Tumour Research

Foundation (DTRF) has suggested a nomogram (analysis of risk factors such as age, gender, and depth of invasion), which gives the score on the risk of the desmoid recurring, is performed before the surgery.^[10] If the score is high (high risk of recurrence), then surgery may not be the best option. Surgery also can be critical for not well encapsulated desmoids within the head and neck region. In such cases, nonsurgical treatments such as radiotherapy, hormonal therapy, chemotherapy, and nonsteroidal antiinflammatory drugs (NSAIDs) have been used to treat desmoid tumors in order to preserve the function of the vital organs.

Few cases of submandibular desmoid fibromatosis have been reported with majority occurring in the pediatric group. Most of these cases opted radical surgical approach with reconstruction.^[11-13] Kutluhan *et al.* reported a case desmoid fibromatosis in a 4-year-old boy presented with mass occupying the mandible completely and extending to the submandibular gland and soft tissues.^[12] Complete

hemimandibulectomy and submandibular gland excision were performed followed by reconstruction with a curved Kirschner wire.^[12] No signs of recurrence were observed during a follow-up period of 18 months. In addition, no limitations in the functions of the jaw, mastication, and swallowing were noted.^[12] We carried out segmental mandibulectomy [Figure 4] in our patient sparing the inferior alveolar nerve and this required no reconstruction of the mandible.

Radiotherapy has been proposed as second option for patients who cannot have surgery or used as adjunct to surgery. Studies have proven that relapse rate has dropped from 59% to 25% when radiotherapy was given postoperatively.^[11] Radiation therapy has to be discussed carefully before considering for small regions such as head and neck because of the radiation effects such as tissue fibrosis or even the development of secondary neoplasm.^[14] With negative resection margin, we opted not to give any adjuvant radiotherapy to our patient.

Besides that, percutaneous cryoablation is also another latest intervention works in the same manner but with use of cold energy such as ice to freeze the tumor.^[15] As radiation therapy has many drawbacks, it should be only considered in cases where surgery can significantly cause functional disabilities in the patient.

For advanced desmoid tumors, several medical treatments such as NSAIDs, antihormonal therapy and chemotherapies, have been suggested if radiotherapy and surgery are potentially harming. Natural history of the disease is crucial for the antihormonal therapy. For example, some studies have indicated that desmoid tumors express nuclear oestrogen receptor-B and the speed of growth of fibromatosis is regulated by female sex hormones.^[16] Therefore, tamoxifen is largely prescribed despite the fact that only a subset of patients respond to antihormonal therapies.^[16] Furthermore, studies have shown that NSAIDs such as indomethacin plays an important role in the pathogenesis of desmoid tumors.^[17] Due to its low side effects, NSAID has become the first line treatment for most advanced desmoid tumors. But to date, there has not been a single case report on the usage of this drug in the treatment of submandibular desmoid fibromatosis.

Even with lack of randomised controlled trial, for life threatening advanced tumors, cytotoxic chemotherapy such as the combination of vinblastine and methotrexate has been prescribed.^[1,13] Othmani *et al.* reported a case of submandibular fibromatosis in a 3-year-old girl.^[13] The tumor appeared clinically aggressive, involving adjacent

tissue and caused bony erosion.^[13] He successfully treated the child with low dose vinblastine and methotrexate and nonmutilating surgical resection.^[13] Complete remission was achieved in 30 months.^[13]

Similar medical treatment of vinblastine and methotrexate was used successfully on a 10-year-old boy with desmoid fibromatosis of the upper lip to avoid mutilating surgery.^[18]

Considering the significant morbidity from surgery, radiotherapy and cytotoxic systemic medications, a period of watchful waiting may be the most appropriate management in asymptomatic patients. A study by Bonvalot *et al.* showed that there was no significant difference between the group that received 'wait and see' and the group that received medical therapy.^[19] The author concluded that conservative policy could be a safe approach to primary and recurrent desmoid fibromatosis, which could avoid unnecessary morbidity from surgery and/or radiation therapy.^[19]

CONCLUSION

At present the optimal treatment of desmoid fibromatosis particularly in the head and neck region needs to be individualized and treated in a multidisciplinary stepwise approach in order to eradicate the disease and reduce the chances of local recurrence. The aim of each individualized therapy should include reducing morbidity and functional loss.

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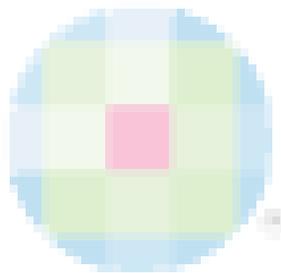
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