

CASE REPORT

A case report of desmoid tumor involving the head and neck region: challenges in diagnosis and treatment

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Desmoid tumor, or aggressive fibromatosis, is a rare condition affecting young adults. Diagnosis is challenging as it has to be established histopathologically by experienced oncopathological experts. Treatment options include surgery, radiation, and other forms of noninvasive modalities like hormonal therapy, low-dose and conventional-dose chemotherapy, non-steroidal anti-inflammatory drugs, tyrosine kinase inhibitors, etc. Each treatment modality has its own advantages and disadvantages. Also, there is a lack of clear guidelines for the management of such tumors, so the treatment-related decisions also become more challenging. Here, we are presenting such a case that was successfully treated using radiation alone with minimal side effects and achieved a good outcome in terms of tumor control, functional preservation, and cosmesis. Hopefully, it will help the medical community in deciding the treatment options and acting as a base for future studies.

Keywords: desmoid tumor, aggressive fibromatosis, radiotherapy, case report, tumor

Introduction

Desmoid (DT), also known as aggressive fibromatosis, is an uncommon condition characterized by infiltrative development and a proclivity for local recurrence but no metastasis. Multifocal origins can occur in the same limb or bodily organ. This uncommon disease accounts for 0.03% of all newly diagnosed cancers (1).

Although DT is harmless in nature, it is locally infiltrative and produces severe morbidity owing to the damage to nearby organs and tissues. We present a case of DT involving the head and neck region that was detected and treated at our hospital. Due to the complicated presentation, difficulties in diagnosis, and lack of standard guidelines backed by robust data, the case was difficult to manage.

Case history

A 22-year-old young adult shows up with a 2-year history of steadily developing right-sided facial edema. It was solid, painless, and fixed to the underlying structures. Teeth in the upper alveolar arch on the right side were found to be loose and deformed. A contrast-enhanced CT scan revealed a large mass epicenter primarily adjacent to the right maxilla measuring 6.5*5.7*5.9 cm (anteroposterior, lateral, and craniocaudal) and eroding adjacent structures such as the hard palate, mandible, lateral pterygoid plate, and reaching up to the infratemporal fossa (**Figure 1**). The probable radiological diagnoses were maxillary cancer, upper gingiva-buccal sulcus carcinoma, or soft tissue sarcoma. However, a biopsy revealed that the patient had severe fibromatosis or a desmoid tumor.

Considering the young age, large tumor size, morbidity associated with an upfront surgical approach, and local aggressiveness of the disease, it was decided by the tumor board that definitive radiation therapy would be the best approach in this case.

Abbreviations: CT, computed tomography; TPS, treatment planning system; AP, anteroposterior; LAT, lateral; CC, craniocaudal.

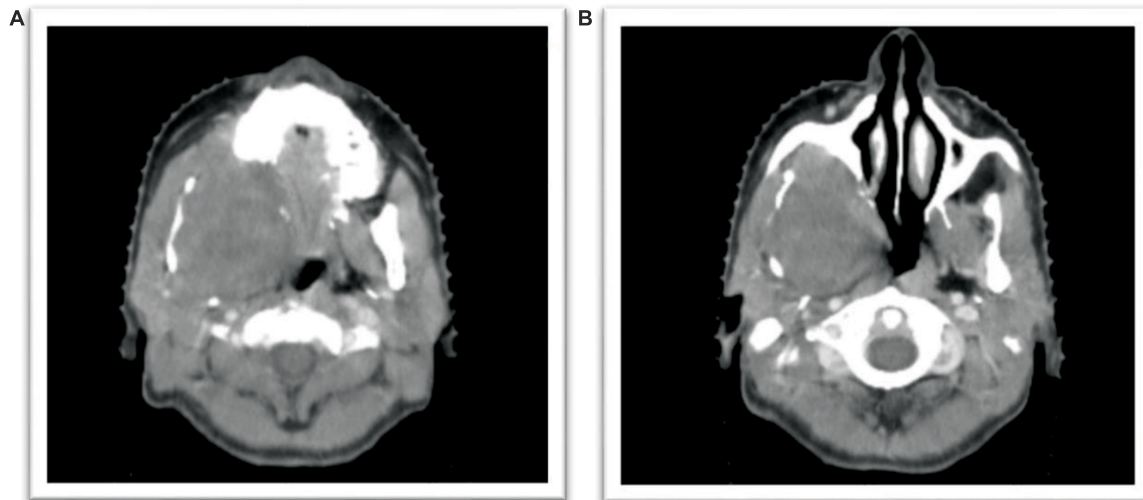


FIGURE 1 | (A,B) showing tumor involving the right maxilla and eroding adjacent structures.

The CT simulation was used with the patient being in a supine position with hands by the side, and a 5-clamp thermoplastic mold was used for positioning and immobilization purposes. The patient was planned using Varian Treatment Planning System Eclipse version 13, using the RapidArc technique with two semi-arcs from 30 to 180 (Figure 2). The daily treatment was delivered using a linear accelerator and 6 MV photons. The total dose given was 5,400 cGy in 30 fractions at a rate of 180 cGy per fraction over 6 weeks. The patient was assessed weekly for various acute toxicities (2) like oral mucositis, radiation dermatitis, and odynophagia during the treatment (Table 1).

On the first follow-up after 30 days from radiotherapy completion, the patient achieved a good clinical response. There was no significant treatment-related morbidity, and

the patient was able to perform routine functions like mastication, deglutition, and phonation. The result was encouraging in terms of cosmesis as well, since the treatment with radiation did not cause any facial disfiguration, pain, or long-term hospitalization that may happen in larger resections.

Discussion

The incidence of DT or aggressive fibromatosis is extremely rare and found in 4–5 cases per million people per year. It primarily affects young adults in their 30–40 s, with a slight female predominance. The location of the DT can range from intra-abdominal to extra-abdominal sites, and around 5–10% of cases are part of familial adenomatous polyposis syndrome. Histologically, these tumors are composed of spindle-shaped cells dispersed throughout the matrix, which is collagenous in nature and devoid of features like hyperchromatic, unusual, and pleomorphic nuclei, which is the hallmark of a typical malignancy (3).

Due to unfavorable location, size of the tumor, and already-caused local destruction, active surveillance was ruled out as an option because it may further complicate the outcome and cause significant mental and physical agony due to treatment-related morbidity to the patient, as observed by Penel et al. (4).

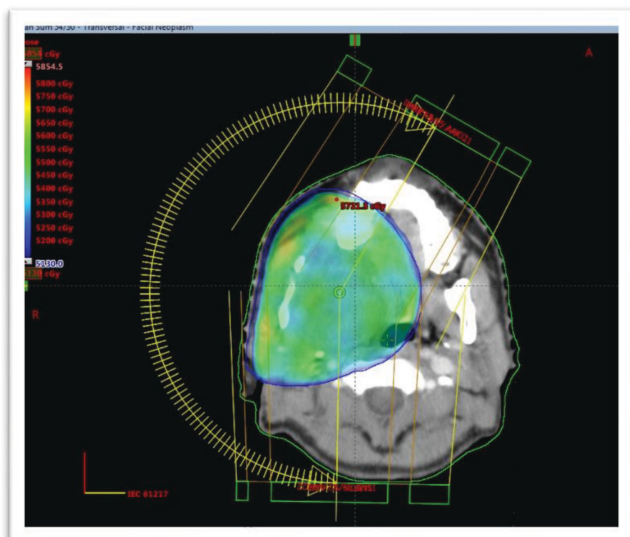


FIGURE 2 | Radiation therapy planned with VMAT (Volumetric Modulated Arc Therapy) technique and its dose coverage.

TABLE 1 | Acute Radiation reactions and status on 1st follow up.

Site	Maximum toxicity during radiotherapy	Toxicity on 30 days post radiotherapy
Skin	Grade II	Grade 0
Mucosal membranes	Grade II	Grade I
Pharynx	Grade I	Grade 0

To avoid treatment-related morbidity and its associated side effects, as the age of the patient is in the early 20 s and being head and neck location, it was decided to consider moderate-dose radiotherapy alone. As compared to surgery followed by radiotherapy, radiotherapy alone was shown to be almost equivalent in terms of recurrence rates (22 vs. 23%, respectively). Even the absolute risk of progressive disease was seen to be 19% with radiation alone versus 29% with surgery alone, with the difference not statistically significant. So, even though surgical modality is the choice of treatment, it is only if it is associated with limited morbidity. When there are chances of significant morbidity or not being able to achieve R0 resection, radiation plays an important role as it gives equivalent local control and progression-free survival (1, 5, 6).

Other treatment options include tyrosine kinase inhibitors (TKIs), non-steroidal anti-inflammatory drugs (NSAIDs), and anti-hormonal therapies. It also includes chemotherapeutic agents in low-dose or conventional-dose regimens typically composed of liposomal doxorubicin, which is usually used in treating soft tissue sarcomas. Perhaps these modalities are experimental in nature, slow acting, and need larger trials for establishing itself as standard of care (7).

Conclusion

Desmoid tumor, also known as aggressive fibromatosis, is an uncommon condition affecting young adults. Diagnosis is challenging as it has to be established histopathologically by experienced oncopathological experts. Treatment options include surgery, radiation, and other modalities like hormonal therapy, low-dose and conventional-dose chemotherapy, NSAIDs, TKIs, etc. Although surgery forms the mainstay of the treatment, various studies using radiation alone also gives the equivalent results. So, the choice of treatment depends on various factors associated with it, like age, general condition, tumor location, expected

morbidity, and outcomes. In the future, it needs larger trials to establish the definitive line of treatment, directly comparing surgery to radiotherapy. Here, we have treated this case successfully using radiation alone with minimal side effects and archived a good outcome in terms of tumor control, functional preservation, and cosmesis.

Summary of the case

1	Patient (gender, age)	Male, 22 years
2	Final diagnosis	Desmoid tumor/aggressive fibromatosis
3	Symptoms	Facial swelling, loose teeth
4	Medications	symptomatic
5	Clinical procedure	Radiotherapy
6	Specialty	Radiation oncology

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