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Desmoid-type fibromatosis of neck masquerading as nerve sheath tumors: two case reports

Surya Kant[1], Bheru Dan Charan[1], Vinay Goel[1], Sumanta Das[2], Saumya Sahu[2], Rajiv Sharma[3], Sachin Borkar[3], Leve Joseph Devaranjan Sebastian[1] and Ajay Garg[1*]

Abstract

Background Desmoid fibromatosis (DF) is an atypical, non-malignant tumor known for its distinctive feature of exhibiting an intermediate biological behavior that falls between benign fibrous lesions and fibrosarcoma. The article aimed to outline the imaging characteristics of desmoid fibromatosis and distinguish it from its similar counterpart, nerve sheath tumors.

Results Here, we present two cases of desmoid fibromatosis whose imaging findings mimic nerve sheath tumors. The patients presented with neck swelling and radiating pain. Both cases were biopsy-proven. Imaging features characteristic of these lesions include elongated shape, split-fat sign, and fascial tail sign with mixed signal intensity on T1WIs and T2WIs depending upon the collagen content of the lesion.

Conclusions MRI is the best diagnostic modality for differentiating fibromatosis from nerve sheath tumors. Surgery continues to be the primary approach for treating these growths, but in some cases, chemoradiation is employed to achieve improved control rates. The presence of intralesional low-intensity bands in T2WIs is a characteristic feature of fibromatosis. Given its local aggressiveness and propensity for recurrence, the use of multiple imaging modalities is essential for providing informed guidance on outcomes and treatment strategies.

Keywords Fibromatosis, Desmoid, Chemoradiation, MRI

Background

Desmoid-type fibromatosis (DF) is an unusual benign locally infiltrative mesenchymal tumor characterized by marked fibroblasts and myofibroblasts proliferation, along with increased production of intercellular collagen within deep musculoaponeurotic structures. It exhibits intermediate biological behavior, falling between benign fibrous lesions and fibrosarcoma [1].

The incidence of this rare benign neoplasm is extremely low, accounting for just 0.03% of all neoplasms [2]. Anatomically, it can be categorized into three types: abdominal wall, extra-abdominal, and intra-abdominal. Head and neck DF cases comprise 10–25% of all cases of extra-abdominal DF [3]. Being a locally aggressive lesion with a tendency to recur, a multimodality approach comprising surgery and chemoradiation can be effective. In this report, we present two cases of DF where imaging findings helped to make a definitive diagnosis. The article aimed to describe the imaging findings of DF and differentiate it from its close mimic nerve sheath tumor.

*Correspondence:
Ajay Garg
ajaygarg@aiims.edu
[1] Department of Neuroimaging & Interventional Neuroradiology, Neurosciences Centre, All India Institute of Medical Sciences, Room No 14, Cathlab Complex, New Delhi 110029, India
[2] Department of Pathology, All India Institute of Medical Sciences, New Delhi, India
[3] Department of Neurosurgery, All India Institute of Medical Sciences, New Delhi, India

Case presentation

Case 1

A 36-year-old male patient presented with a gradually progressing swelling on the left side of his neck that had developed insidiously over the past four years. He also reported sharp shooting pains and gradually losing strength in his left upper limb. Interestingly, he did not experience any numbness, tingling, sensory loss, difficulty swallowing, or voice hoarseness.

On examination, weakness in the left upper limb was noted, but no sensory loss was seen. The neck mass was large, firm, and non-tender, extending from the angle of the mandible to the clavicle on the left side of the neck.

Magnetic Resonance Imaging (MRI) revealed a large, well-defined multilobulated lesion in the region of the left brachial plexus, extending from the base of the skull down to the T2 vertebra on the left side of the neck. It involved the paraspinal area, left prevertebral region and extended superiorly into the left parapharyngeal space. It displaced the left carotid space anteriorly and pushed the trachea and esophagus toward the right side (Fig. 1). The lesion encased the left brachial plexus from the trunk onward and displaced the left subclavian artery and vein anteriorly. The lesion was isointense on T1WIs, hyperintense on T2WIs, and STIR (Short-TI Inversion Recovery) images, and displayed intense enhancement following contrast administration. Multiple hypointense bands are observed on T2WIs. Diffusion-weighted imaging did not show any restricted diffusion, and there was no presence of an eccentric dot sign. Based on these radiological findings, desmoid-type fibromatosis was diagnosed (Fig. 1).

Histopathological analysis of the tissue sections revealed a spindle cell tumor arranged in a fascicular pattern within a myxoid and collagenous stroma. Thin-walled dilated blood vessels were interspersed throughout the tissue (Fig. 2). There was slight nuclear atypia, but no mitotic activity or tissue necrosis was observed. Additionally, scattered lymphocytes and mast cells were identified within the specimen. Immunohistochemistry revealed nuclear positivity for beta-catenin in the tumor cells. However, the tumor cells tested negative for CD34, SMA, and S100. Based on these findings, the final

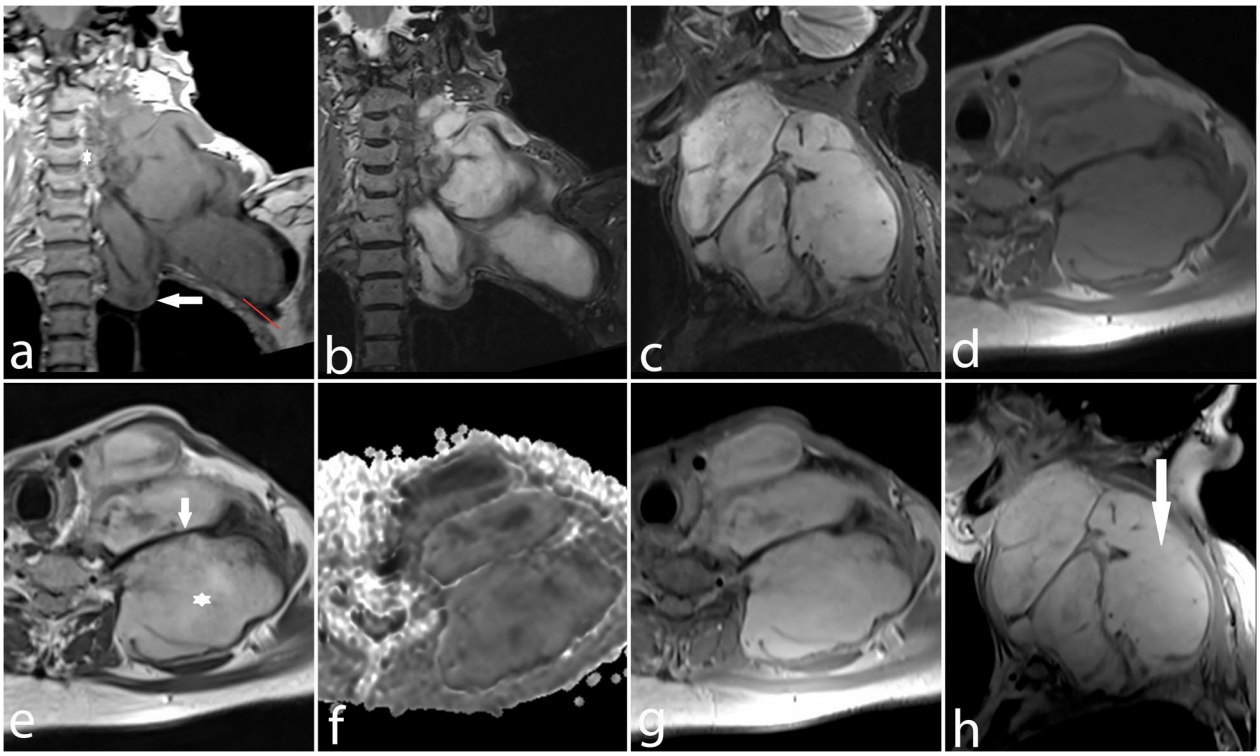

Fig. 1 A 36-year-old male patient presented with a gradually developing swelling on the left side of his neck that had been ongoing for the past four years. MRI findings revealed the following: Coronal T1WI **a** shows a large, well-defined, multilobulated mass lesion with an isointense appearance on the left side of the neck. This mass extends along the left brachial plexus (red line) within the fascial planes, medially entering the left neural foramina and extending inferiorly into the mediastinum (indicated by white arrow). In coronal **b** and sagittal **c** STIR (Short-TI Inversion Recovery) images, the mass appears hyperintense, while it appears isointense in axial T1WI **d**. The mass displays hyperintensity (indicated by *) on axial T2WI (**e**), and multiple hypointense bands (indicated arrow in **e**) are seen within it. Notably, DWI maps **f** show no diffusion restriction within the mass. Post-contrast axial (**g**) and sagittal (**h**) T1WIs reveal heterogeneous intense enhancement (arrow in **h**) of the mass, with a few non-enhancing areas within the lesion

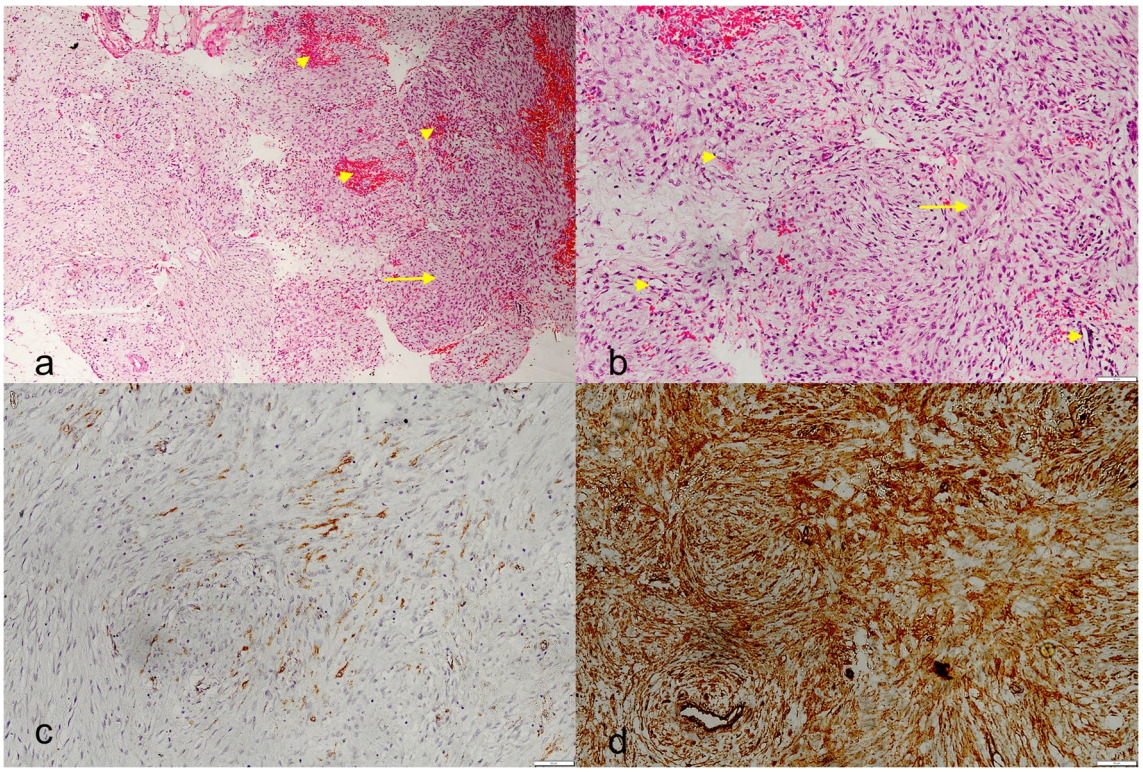

Fig. 2 In histopathological examinations, the following observations are made: At 4X magnification in the Hematoxylin and Eosin (H&E)-stained sections (**a**), there are fascicles of spindle-shaped cells (indicated by the arrow), along with areas showing microhemorrhages (indicated by the arrowhead). At 10X magnification in the H&E-stained sections (**b**), the spindle cells are visible with vesicular nuclei and pinpoint nucleoli (arrow) and nearby thin-walled blood vessels (arrowheads). Immunohistochemistry for Smooth Muscle Actin (SMA) reveals focal positivity, presenting a 'tram-track' pattern (**c**). Immunohistochemistry for Beta-catenin exhibits strong nuclear staining (**d**)

diagnosis of desmoid-type fibromatosis involving the left brachial plexus was confirmed (Fig. 2).

Case 2

A 22-year-old female patient complained of intermittent neck pain, numbness, and weakness in her left shoulder that had persisted for a year. She had no history of difficulties with swallowing, breathing, or hoarseness of voice.

On examination, weakness was evident in her left shoulder and left elbow and sensory loss was noted in the left C3, C4, and C5 dermatomes.

MRI revealed a well-defined lobulated multicompartmental mass in the region of the left brachial plexus. The mass extended from C2 to C6 vertebral level and occupied the left paraspinal, prevertebral, and parapharyngeal spaces, pushing the trachea and esophagus to the right side and displacing the left carotid sheath anterolaterally (Fig. 3). Additionally, the lesion extended into the left neural foramen at C2–C6 disc levels, leading to focal narrowing of the left vertebral artery due to compression. The lesion appeared isointense on T1WIs, heterogeneously hyperintense with multiple hypointense bands on T2WIs, hyperintense on STIR WIs images, and had intense enhancement following contrast administration.

Further examination through CT angiography confirmed that the lesion extended into the left neural foramen at the C2-C6 level, causing focal compression and narrowing of the left vertebral artery. Based on these radiological findings, desmoid-type fibromatosis was diagnosed (Fig. 3).

Surgical cervical exploration and subtotal decompression of the tumor were done. The tumor had a stony-hard texture, appearing grayish-yellow, and was firmly adhered to nearby nerves. The excised tumor was subsequently sent for histopathological examination.

Histopathological analysis of the sections revealed a spindle cell tumor, partially circumscribed but with infiltrative margins (Fig. 4). The tumor cells were arranged in interlacing fascicles and displayed mild oval to spindle-shaped nuclei with minimal atypia. No areas of necrosis were observed. Three lymph nodes were identified, all exhibiting reactive lymphoid hyperplasia.

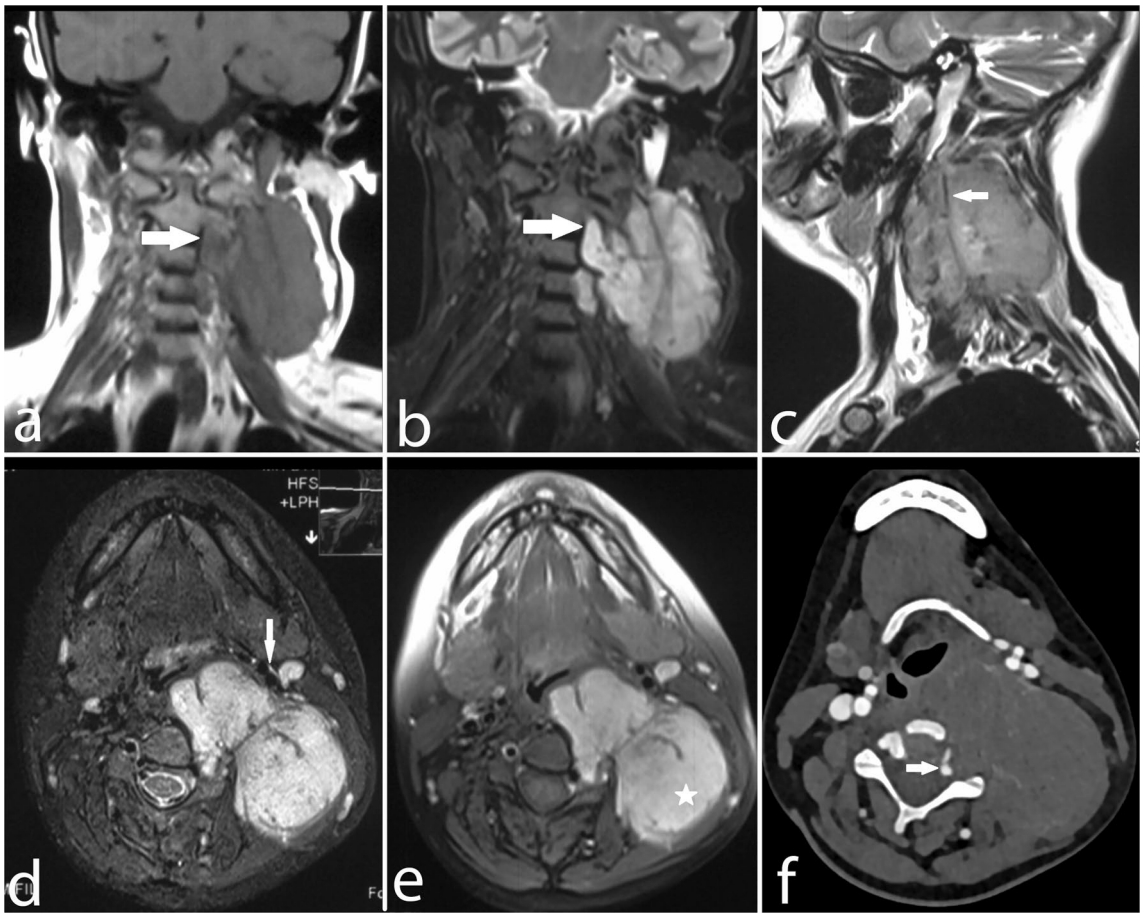

Fig. 3 A 22-year-old female patient reported experiencing occasional neck pain, along with episodes of numbness and weakness in her left shoulder that had persisted for a year. MRI findings reveal the following: Coronal T1WI **a** shows a well-defined multilobulated isointense lesion on the left side of the neck extending along the left brachial plexus within the facial planes and medially within the left neural foramina (indicated by arrow in **a** and **b**). The mass is hyperintense in coronal STIR image (**b**) and heterogeneously hyperintense with multiple hypointense bands (arrow in **c**) in sagittal T2WI (**c**). The mass pushes the trachea and esophagus toward the right side, shifts the carotid sheath anterolaterally (arrow in **d**), and extends into the left neural foramen (**d**). Post-contrast axial T1WI e shows an avid enhancing (* in **e**) lesion with a few non-enhancing areas. In the axial CT angiography image (**f**), the left neural foramina is expanded, accompanied by narrowing and medial displacement of the left vertebral artery (horizontal arrow in **f**).

Immunohistochemical analysis indicated that the tumor cells were positive for SMA (focal), ATRX (retained), and Beta-catenin (diffuse nuclear), while testing negative for S100, EMA, CD34, P53, and SOX10. The MIB-1 labeling index was 3%. These features were consistent with a diagnosis of desmoid-type fibromatosis, characterized by a hypercellular pattern (Fig. 4).

Discussion

Desmoid fibromatosis of the head and neck is a rare benign mesenchymal tumor with an annual incidence of 2–4 cases per million individuals [2]. Its origins are multifactorial, potentially influenced by genetic, traumatic, and hormonal factors [4, 5]. Despite its benign nature, it tends to be locally aggressive and infiltrative. While it can develop in various fibrous tissues throughout the body, it most often occurs in the abdomen, constituting nearly two-thirds of all DF cases [6]. Head and neck DF comprises approximately 10–25% of all extra-abdominal cases [3]. The anterolateral aspect of the neck is a common site for these tumors.

Notably, their significance in the neck arises from their proximity to vital neurovascular structures, making surgical intervention particularly challenging. Consequently, a multimodal approach to management is often necessary to control the disease.

DF typically presents as a painless mass, though, as observed in our cases, pain is common. Most clinical symptoms arise from mass- effects over adjacent neural and vascular structures. For instance, compression over

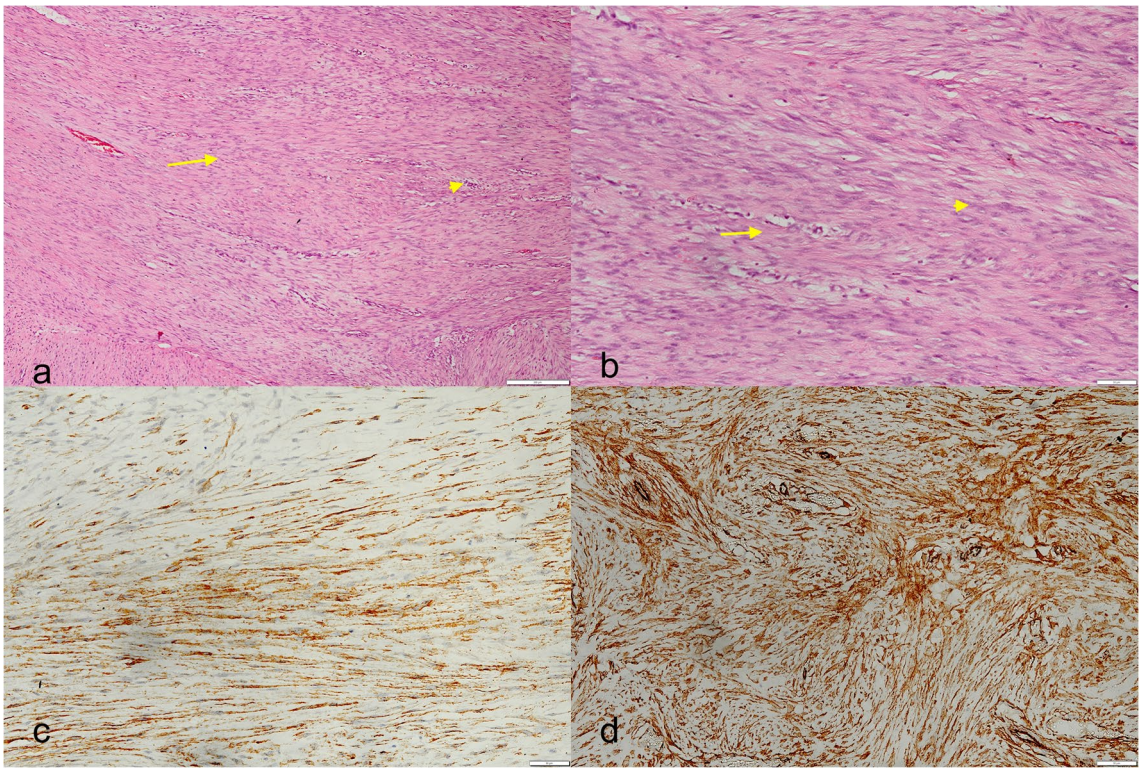

Fig. 4 The histopathological examination reveals that at 4X magnification in the H&E-stained sections (**a**), there are long, sweeping fascicles of elongated spindle cells within a collagenous stroma (indicated by the arrow). Perivascular edema is also observed (indicated by the arrowhead). At 10X magnification in the H&E-stained sections (**b**), fascicles of spindle cells are visible within the collagenous stroma (arrow), featuring vesicular nuclei with pinpoint nucleoli (arrowhead). Immunohistochemistry for Smooth Muscle Actin (SMA) exhibits staining in a 'tram-track' pattern (**c**). Immunohistochemistry for Beta-catenin displays strong nuclear staining (**d**)

the brachial plexus can result in symptoms such as tingling, paranesthesia, numbness, and weakness in the hand muscles. As seen in our first case, shooting pains radiating from shoulder to hand have been previously reported [6].

The first step in diagnosing these tumors typically involves imaging, especially CT and MR imaging. On CT scans, DFs appear as hyperattenuating masses with avid enhancement on post-contrast CT scans [7]. CT imaging can provide valuable insights into bone erosions and scalloping.

MR imaging is considered the best imaging modality for a more comprehensive evaluation and staging. These lesions often occupy intermuscular locations with a surrounding rim of fat termed the 'split-fat sign.' Another characteristic feature is linear extension along fascial planes, known as the 'Fascial tail sign' [8]. This growth pattern gives these lesions their characteristic 'elongated shape', as observed in our case [9]. While the appearance of DF on MRI can vary depending on the concentration of collagen and fibrous components within the lesion, they generally display intermediate signal intensity on T1WIs (similar to muscles) and T2WIs (lower than fat but higher than adjacent muscles) [10, 11]. Fat-suppressed T2WIs MR sequences often reveal higher signal intensity within these lesions, as seen in our cases. Regions of low signal intensity bands in DF correspond to higher collagen content within the lesion [8].

Post-contrast images typically reveal moderate to marked heterogeneous enhancement of these lesions. Due to their tendency to infiltrate vital neck structures such as the trachea, brachial plexus, and vessels, surgical resection is often limited. Goldstein et al. [12] described a similar case of a desmoid tumor in the neck in a 62-year-old male. Eksi et al. [13] reviewed the literature in their article on spinal desmoid tumors.

The differential diagnosis for DF includes neurogenic tumors, fibrosarcoma, solitary fibrous tumors, and rhabdomyosarcoma. Diffusion-weighted imaging may be useful to differentiate malignant soft tissue tumors of the neck from DF, as the latter typically exhibits a high apparent diffusion coefficient value [14].

Due to the challenges of achieving a wide margin during surgical excision and a high local recurrence rate of

30–40%, radiation therapy is often combined with surgery to achieve better control rates [2, 15].

Conclusions

Desmoid fibromatosis in the head and neck regions have distinctive imaging such as elongated shape, the presence of the split-fat sign, and the fascial tail sign. These tumors typically exhibit low-intensity bands on T2WIs within the lesion. While surgery remains the primary approach for treating these lesions, there is a growing exploration of chemoradiation as a means to achieve improved control rates.

Abbreviations

DF Desmoid fibromatosis
MRI Magnetic resonance imaging
STIR Short tau inversion recovery

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Not applicable.

Author contributions

SK, BDC, VG, SD, SS, RS, AG, and SLJD contributed to acquisition, analysis, conception, design, and drafting of the work. SB, along with SK, BDC, VG, SD, SS, RS, AG, and SLJD contributed to the final draft, revisions, upload, and submission of final revised work. All authors have agreed both to be personally accountable for own contributions and ensured that questions related to the accuracy or integrity of any part of the work, even ones in which one was not personally involved, are appropriately investigated, resolved, and the resolution documented in the literature.

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Availability of data and materials

Yes, on request.

Declarations

Ethics approval and consent to participate

This work has been approved by Institute Ethical Committee (IEC). Retrospective, consent waiver

Consent for publication

Consent for publication has been obtained from the patient in writing, however, their identity is not disclosed. All the authors have approved submitting the manuscript to your esteemed journal. On behalf of all the contributors, I will act as guarantor and correspond with the journal from this point onward.

Competing interests

The authors declare that they have no competing interests.

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