Desmoid tumor: Multifarious and challenging. The role of MRI

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Learning objectives

To illustrate the MRI appearance in different types of desmoid tumors, describing imaging characteristics that can contribute to specific diagnosis.

Background

• Definition

Desmoid tumors (DTs), also known as fibromatoses, are rare, non-malignant neoplasms of mesenchymal origin, demonstrating aggressive fibroblastic proliferation. The lesions are comprised of benign fibrous tissue with elongated or spindle-shaped cells adjacent to collagen. Desmoids usually do not exhibit signs of metastasis, but they commonly have local infiltration and recurrence tendency (a recurrence rate of 65% has been reported).

• Classification

DTs have been classified by location as superficial (palmar, plantar, penile, infantile digital fibromatosis) or deep (extra-abdominal, abdominal, intra-abdominal).

The tumors fall into two categories: Sporadic type, usually presenting as extra-abdominal/abdominal-wall mass and familial type, mainly intra-abdominal and often associated with adenomatous polyposis (most commonly with Gardner syndrome and Turcot syndrome).

Aggressive fibromatosis usually occurs in the deep soft tissues and is characterized by proliferation of fibrous tissue that infiltrates adjacent structures. This type of fibromatosis is found to have greater tendency to recur locally, especially postoperatively, compared to well-circumscribed DTs.

• Etiology

The etiology of DTs is not yet evident and is believed to be multifactorial. Genetic, endocrine, and physical factors play an important part in the tumors’ appearance. An association with a germline mutation in the adenomatous polyposis coli (APC) gene, related with hereditary polyposis syndromes, has been reported. On the other hand, sporadic DTs are commonly associated with somatic mutations of the codons 41, 45 of
exon 3 of the beta-catenin gene (CTNNB1). There is a reported connection between DTs and multiple, variable, diseases at the molecular level (fig. 1).

Fig. 1: Graphical network of the top 20 diseases related to Desmoid Tumor via text searches within MalaCards or GeneCards/GeneDecks gene sharing.

References: Weizmann Institute of Science - www.malacards.org

Besides polyposis syndromes, a relation between desmoids and other likely pathogenetic factors, such as history of abdominal or pelvic surgery, trauma, estrogen therapy, hormonal imbalance and pregnancy, has been described.

• Epidemiologic features

1. Frequency of Occurrence
   » Superficial fibromatosis: 1.5% of benign soft tissue tumors.
   » Deep fibromatosis (adults): 5% of benign soft tissue tumors (with an incidence of 2-4 per million per year).

2. Age at Presentation, Gender Ratio (M/F)
   » Superficial fibromatosis: from 17 to 65 years (mean: 41 years), 2/1.
   » Deep fibromatosis (adults): from 13 to 60 years (mean: 34 years), 1/1.4 to 1/2.
» Deep fibromatosis (children): from 6 months to 8 years (mostly in first 2 years), predominantly in males.

3. **Location in soft tissue**
   
   » Superficial fibromatosis: Foot and ankle > hand and wrist > trunk.

   » Deep fibromatosis (adults):


   b. Abdominal wall desmoids: rectus, oblique and transversus muscles, musculoaponeurotic structures and/or fascia.

   c. Extra-abdominal desmoids: chest wall, mediastinum, pelvic wall, paraspinal space, extremities (lower extremity > foot and ankle > hand and wrist > upper extremity), head and neck.

   » Deep fibromatosis (children): Head and neck (tongue, mandible, maxilla, mastoid), trunk, proximal extremities. *Intra-abdominal desmoids in children are uncommon.*

**Microscopic appearance - Stages of evolution**

Histologically, DTs are comprised of fibroblasts and myofibroblasts characterized by tapered wispy cytoplasm, elongated vesicular nuclei and multiple small nucleoli. The cells, usually elongated, stellate or bland spindled cells, are positioned in a linear array, encircled and set apart from each other by collagen (fig. 2).
Fig. 2: Microscopic images of DTs A: Cellular fibrous proliferation in elongated fascicles, with numerous slit-like vessels, characteristic of desmoid fibromatosis. B: Fibroblasts having spindled, dense, wavy nuclei and minimal cytoplasm.

References: 2003-2012, PathologyOutlines.com, Inc.

Three stages of evolution of DTs have been described (Vandevenne et al):

1. **First stage:** Mostly cellular lesions - fewer areas of hyalinized collagen.
2. **Second stage:** Increasing amount of collagen deposition in the central and peripheral areas of the tumor.
3. **Third stage:** Increase in the fibrous composition - Decrease in cellularity and water content.

- **Imaging - The role of Magnetic Resonance Imaging (MRI)**

# Imaging procedures: Radiographic study constitutes a significant part in the diagnostic process of DTs.

Plain radiographs may indicate the presence of a soft-tissue mass, but in most cases they cannot detect the lesions. Ultrasound can depict a hypoechoic mass and roughly describe its contour and/or infiltrative behavior (fig. 3). Computed tomography (CT) usually demonstrates a hypo/isodense lesion (homogeneous or inhomogeneous), showing moderate to significant contrast enhancement (fig. 4). MRI has been proven to be the preferable radiologic examination for DTs' detection and characterization.
Method of choice: Among the main imaging techniques, MRI provides the optimum method for evaluating tumor expanse, mass effect and possible resectability, especially in case of extra-abdominal lesions. Having the advantage of high-resolution distinction of soft tissues compared to other radiological modalities, it can demonstrate any invasion into adjoining neurovascular structures. Cross-sectional imaging with CT may be useful for clarifying the tumor's relation to surrounding tissues in order to determine the therapeutic approach.

The role of nuclear medicine techniques in the assessment of DTs is yet to be designated.

- Differential diagnosis list

  # Superficial fibromatosis:
  
  Post-traumatic scar involving tendon or aponeurosis
  
  Giant cell tumor of tendon sheath
  
  Tendon sheath fibroma
  
  Granuloma
  
  Gout

  # Deep fibromatosis:
  
  Liposarcoma, leiomyosarcoma, fibrosarcoma, rhabdomyosarcoma, Kaposi sarcoma, epithelioid sarcoma, clear-cell sarcoma, synovial sarcoma
  
  Malignant fibrous histiocytoma
  
  Malignant schwannoma
  
  Malignant granular cell tumor
  
  Nodular fasciitis
  
  Neurofibroma
  
  Giant cell tumor of tendon sheath, tendon sheath fibroma
  
  Granular cell tumor
  
  Leiomyoma
  
  Proliferative fasciitis/myositis
Images for this section:

**Fig. 3:** Ultrasound image of abdominal desmoid tumor: Sagittal view demonstrates a large, hypoechoic, inhomogeneous lesion with relatively well-demarcated margins, although not clearly delineated from surrounding tissues at the lateral edges.
**Fig. 4:** Contrast-enhanced computed tomography scan demonstrating a desmoid tumor originating from the abdominis transversus and internal oblique muscle fascia, with an inhomogeneous formation, showing moderate enhancement (arrow indicates tumor).
Findings and procedure details

• MRI examination: Procedure and findings

1. Protocol:

A standard MRI protocol for DTs should include the required sequences for soft-tissue lesions imaging:

# T1-weighted images (T1WI) in at least two orthogonal planes (axial and coronal/sagittal): Apart from conventional spin-echo, a gradient-echo T1-weighted technique may also be used.

# T2-weighted images (T2WI) obtained in at least two orthogonal planes.

# Fast spin-echo T2WI with fat saturation or other fat-suppression technique, like STIR (Short Tau Inversion Recovery), in the axial and coronal/sagittal plane.

# Pre- and post-contrast T1WI with fat saturation, in the axial and/or coronal and sagittal plane.

# Dynamic contrast-enhanced T1-weighted sequences (to depict progressive and predominantly delayed enhancement).

2. Findings:

A. Lesion morphology: Tumors can have discrete, well-defined margins (49%-54%) or irregular contour/invasive margins (46%-51%), sometimes infiltrating along the fascial planes. They can be lobulated or regular in shape.

B. Signal intensity and contrast enhancement: These features on MRI depend, to a significant extent, on the lesion's chronicity (corresponding to its stage of evolution) and aggressiveness.

# Signal intensity: T1WI most frequently demonstrate lesions with low to intermediate signal intensity (hypo/iso-intense to muscle), either homogeneous or heterogeneous. On T2WI and STIR sequences, lesions show variable, customarily intermediate to high, signal. High signal intensity on T2WI usually reflects high cellularity of the tumor, correlating with actively growing or recurrent tumor. Inversely, low T2 signal intensity can
indicate hypocellularity and is often related with lesions containing significant amounts of collagen.

*Band-like low-intensity regions* often depicted on T2WI and sometimes on T1WI, correspond to the presence of collagen bundles inside the lesion.

**Enhancement:** Varying types and levels of enhancement after paramagnetic contrast agent administration have been described, but DTs are mainly characterized by *marked gradual enhancement*, better depicted throughout the dynamic phases. The degree of homogeneity varies, depending on the deposition/distribution of spindle cells, collagen, and myxoid component within the lesion. Following contrast medium administration, the low-intensity band-like regions within the lesions demonstrate lack of enhancement. Relation between the pattern or degree of enhancement and tumor recurrence has not been determined.

### 3. Issues concerning the differential diagnosis:

Although MRI features are often highly specific for DT diagnosis, problems in the differential diagnosis emerge regularly. The main difficulty regards similarities in MRI appearance among aggressive fibromatosis and soft-tissue sarcoma, such as *inhomogeneous signal intensity on T2WI*, a common feature mostly depending on the fibrous portion within the lesion (fig. 5).
Fig. 5: a & d: Axial and sagittal T1-weighted spin echo images of the right thigh depict a well-defined, heterogeneous mass, showing isointensity to adjacent muscle, with irregular septum. b & e: Axial and sagittal T2-weighted spin echo images show a multi-lobulated hyperintense mass involving most of the flexor muscles. c & f: Mass enhances in a heterogeneous manner after IV paramagnetic contrast medium administration. Biopsy-histopathology demonstrated unclassified high-grade sarcoma.

References: Kanamori M. et al, CD99-positive soft tissue sarcoma with chromosomal translocation between 1 and 16 and inversion of chromosome 5, Oncology letters June 2012, Volume 3 - Issue 6; Pages: 1213-1215

An important observation is that sarcomas tend to exert pressure on adjoining structures while expanding centrifugally, rather than infiltrate into them. Another problem in differentiating DTs from malignant tumors is that they can present with common imaging characteristics, primarily heterogeneity, ill-demarcated margins and encapsulation of adjacent neurovascular structures. It should be noted, though, that it is improbable for DTs to show signs of necrosis or cystic degeneration, like malignant lesions do.

- Description of MRI studies in various cases of deep desmoid-type fibromatosis

1. Presentation:
Six patients (four female/two male, age range: 14-77 years), most presenting as idiopathic cases, either with palpable extra-abdominal/abdominal-wall mass (four subjects) or in terms of post-surgical follow-up of intra-abdominal desmoid lesion (one subject) and one having a history of Gardner's syndrome (familial case), all underwent MR imaging at 1.5T, in a three-year period.

2. Procedures:

# Three of the patients were subjected to abdominal and pelvic MRI, and two underwent abdominal MRI monitoring. MR protocol included: pre-contrast and post-contrast T1WI, T2WI, fat-saturated T2-weighted HASTE (Half-Fourier-Acquired Single-shot Turbo spin Echo), STIR and Dual Fast Field Echo (DualFFE) sequences, 3D-T1WI with fat suppression, Dynamic-enhanced 3D-T1WI with fat suppression and Single-shot Diffusion-weighted images (SSh-DWI), acquired in axial, coronal and sagittal planes.

Concerning the familial case, additional Steady-state b-GRE (balanced gradient echo) and 3D steady-state b-GRE acquisitions, were obtained in the axial and coronal plane.

# One patient was submitted to MRI of the knee, using a four-element phased array coil. MR protocol incorporated pre-contrast and post-contrast T1WI and STIR sequences obtained in the axial, coronal and sagittal plane, as well as axial T2WI, SSh-DWI and fat-saturated Dynamic-enhanced 3D-T1WI.

3. Sites of involvement:

# Sporadic cases: rectus abdominis sheath (3 subjects), psoas muscle (1 subject), posterolateral compartment of the knee (1 subject).

# Familial case (Gardner's syndrome): mesenteric and pelvic masses.

4. MRI appearance:

» On T1WI, masses produced intermediate to low signal, compared to adjacent muscle. T2-weighted, STIR and fat-saturated HASTE sequences revealed increased signal intensity in five subjects, with predominantly inhomogeneous formation - coexistence of low-intensity areas (fig. 6,7). In one case, lesions with a lesser degree of cellularity and increased collagen content, as verified histologically, showed T2 hypointensity (fig. 8).

» Notable enhancement was present in all cases, as displayed on fat-suppressed post-contrast T1WI and 3D-T1WI, gradually increasing throughout the dynamic phases (fig. 9-10).
Indiscrete lesion margins and/or adjacent tissue infiltration in two patients implied local invasion (fig. 11, 14), while the rectus abdominis sheath masses in three subjects showed relatively distinct margins (fig. 6, 8). Signs of postoperative recurrence were denoted in two of the studies (fig. 12-13, 14).

Morphology of the lesions, signal intensity on T1WI and T2WI, pattern of enhancement and infiltrating behavior, represent specific MRI characteristics which indicated a diagnosis of DT or suggested recurrence of pre-existing DT.

Histopathological confirmation of desmoid tumor was provided in all cases.

Images for this section:

**Fig. 6:** a & b: 41-year-old female with palpable left upper abdominal wall mass and moderate pain. Well-margined lesion in the left rectus abdominis sheath (arrow), appearing isointense to adjacent muscle on fat-suppressed 3D T1-weighted acquisitions (a) and hyperintense, slightly inhomogeneous, on T2WI (b). c & d: Female patient, 14 years old, presenting with palpable left lower abdominal wall mass. MRI demonstrated a mass lying within the left rectus abdominis sheath, having relatively distinct margins (rectangular outline). On T1WI (c), the lesion shows signal intensity similar to adjacent muscle, while on T2WI (d), it appears markedly heterogeneous, producing increased signal compared to muscle, with coexisting band-like low-intensity regions in the medial half of it.
Fig. 7: 77-year-old female with palpable mass in the posterolateral compartment of the right knee-lower thigh. MRI depicted a sizeable lobular mass (measuring about 6.5 cm in maximum diameter) along the short head of biceps femoris muscle. On T1WI in the axial (a) and sagittal (b) plane, the mass appears isointense to adjacent muscles, whereas STIR axial (c) and sagittal (d) images depict marked, inhomogeneous, hyperintensity of the lesion (arrows in a & c and oval outlines in b & d, indicate the mass). Observe the presence of chord-like areas within the lesion, more prominent on STIR images (c & d).
Fig. 8: 39-year-old female, with persistent epigastric pain. MRI revealed a well-circumscribed, mildly lobulated lesion, localized in the superior part of the left rectus abdominis sheath at the level of the ipsilateral costal margin, inducing an "imprint" on the anterior surface of the left hepatic lobe (rectangular outline). Axial fat-suppressed 3D-T1WI (a) demonstrates an inhomogeneous, isointense to muscle, lesion. On axial T2WI (b), it appears heterogeneously hypointense compared to muscle (corresponding to hypocellularity and abundant amounts of collagen, as histopathology revealed).
**Fig. 9:** a & b: 14-year-old female with left rectus abdominis sheath mass [see fig. 6; c & d]. Axial fat-suppressed T1WI (a) depicts a heterogeneous, relatively isointense to muscle lesion, containing hypointense band-like areas in the center and medial part (oval outline). Notable inhomogeneous enhancement is observed on axial post-contrast T1WI with fat saturation (b), with the low-intensity areas lacking enhancement. c & d: 77-year-old female with palpable mass in the posterolateral compartment of the right knee [see fig. 7]. A sizeable lobular mass (oval outline), extends along the short head of biceps femoris muscle at the level of the patellar base, coursing cephalically, effacing the space between the biceps femoris and the posterior inferior edge of the vastus lateralis and vastus intermedius muscles and obliterating the lateral head of the gastrocnemius muscle, as well. Coronal T1WI (c) exhibits an isointense lesion compared to normal muscle, with embodied chord-like low-intensity regions. On coronal fat-suppressed post-contrast T1WI (d), prominent inhomogeneous enhancement is demonstrated, with chord-like low-intensity regions showing minimum enhancement.
Fig. 10: 41-year-old female with mass in the left rectus abdominis sheath [see fig. 6; a & b]. On axial fat-suppressed 3D-T1WI prior to contrast medium administration (a), the lesion appears isointense compared to adjacent muscle (rectangular outline), showing notable, rather homogeneous enhancement (arrow) on post-contrast acquisitions (b).
**Fig. 11:** 28-year-old male with Gardner’s syndrome, underwent follow-up MRI after total colectomy. Coronal HASTE sequence with fat suppression (a) depicts an ill-margined hyperintense mesenteric mass, extending caudally to the pelvic inlet, surrounding mesenteric arterial and venous branches (rectangular outline). Axial steady-state gradient echo acquisition of the pelvis (b) reveals another hyperintense lesion of indistinct margins occupying the presacral space, extending along the sacral nerves into the sacral foramina, bilaterally (rectangular outline). On axial post-contrast fat-saturated 3D-T1WI (c & d), both lesions show marked, rather inhomogeneous, enhancement (oval & rectangular outline, respectively).
Fig. 12: Male patient, 45 years old, underwent control MRI study six months after resection of histologically proven desmoid tumor originating from the right psoas muscle. Axial in-phase image of dual FFE acquisition (a) shows postoperative appearance of the right psoas (blue oval outline) with deformation and relative size reduction, compared to the contralateral-left psoas muscle (green oval outline). Axial fat-suppressed 3D-T1WI (b) demonstrates confluent lesions of node-like morphology at the lateral edge of the right psoas, the adjacent perirenal fat and along the anterior layer of the thoracolumbar and transversalis fascia, ipsilaterally (arrows). These node-like lesions show marked inhomogeneous, mainly peripheral, enhancement, on axial and coronal post-contrast fat-suppressed 3D-T1WI (c & d; rectangular outlines). The MRI findings are indicative of residual desmoid tumor masses/ recurrence in the region of resection.
Fig. 13: Same patient, with resected psoas muscle desmoid tumor [see fig. 12], was subjected to MRI monitoring, three months later. Post-contrast fat-saturated 3D-T1WI, acquired in the axial (a,b,c) and coronal (d,e) plane, exhibit notable heterogeneous enhancement of the node-like lesions, which appear moderately increased in size (oval outlines in the axial images/ arrow & triangular outline in the coronal images, respectively). MRI findings are suggestive of recurrence.
Fig. 14: 77-year-old female patient with soft-tissue mass in the posterolateral compartment of the right lower thigh [see fig. 7 and fig 9; c & d]. Pre-surgical axial T1-weighted (a), STIR (b) and post-contrast fat-suppressed T1-weighted (c) images, depict the lesion (asterisk) expanding along the short head of biceps femoris muscle, destroying the delineation of surrounding structures and effacing the posterior inferior edge of the vastus lateralis - vastus intermedius and the superior aspect of the lateral head of the gastrocnemius muscle. The mass extends towards the iliotibial band and the subcutaneous tissues, laterally. Signal intensity and enhancement pattern (hypointense on T1WI and inhomogeneously hyperintense on STIR, with marked heterogeneous enhancement) as well as locally aggressive behavior, are suggestive of fibromatosis. The mass was excised and desmoid tumor diagnosis was verified histopathologically. Post-surgical axial T1-weighted (d), STIR (e) and post-contrast fat-suppressed T1-weighted (f) images, six months later, depict a mass within the resection region, along the short head of biceps femoris muscle (about 4.5cm in anteroposterior diameter), with extension into surrounding muscles and neurovascular structures (oval outline). The lesion demonstrates similar MRI characteristics on d,e & f images to the pre-operative images (a,b & c, respectively), appearing less inhomogeneous, though. The findings are indicative of desmoid tumor recurrence.
Conclusion

MRI is the key imaging technique for initial preoperative diagnosis and post-therapeutic follow-up of desmoid tumors (a quite diverse entity), providing accuracy in assessing tumor growth and infiltration, as well as evaluating the response to treatment by estimating changes in size and signal intensity, better displayed on T2-weighted images.

To summarize, characteristic location along with specific MRI signal intensity and contrast enhancement features may lead to determined diagnosis of DTs. Histopathological correlation is required at all times, though.

Personal information

References


