Retrospective Study

Clinical and imaging features of desmoid tumors of the extremities

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 Abstract

 Background
 Desmoid fibroma is a rare soft tissue tumor originating from the aponeurosis, fascia, and muscle, and it is also known as aponeurotic fibroma, invasive fibroma, or ligamentous fibroma.

 Aim
 To investigate the clinical and imaging features of desmoid tumors of the extremities.

 Methods
 Thirteen patients with desmoid fibroma of the extremities admitted to our hospital from October 2016 to March 2021 were included. All patients underwent computed tomography (CT), magnetic resonance imaging (MRI), and pathological examination of the lesion. Data on the diameter and distribution of the lesion, the relationship between the lesion morphology and surrounding structures, MRI and CT findings, and pathological features were statistically analyzed.

 Results
 The lesion diameter ranged from 1.7 to 8.9 cm, with an average of 5.35 ± 2.39 cm. All lesions were located in the deep muscular space, with the left and right forearm each accounting for 23.08% of cases. Among the 13 patients with desmoid fibroma of the extremities, the lesions were “patchy” in 1, irregular in 10, and quasi-round in 2. The boundary between the lesion and surrounding soft tissue was blurred in 10 cases, and the focus infiltrated along the tissue space and invaded the adjacent structures. Furthermore, the edge of the lesion showed “beard-like” infiltration in 2 cases; bone resorption and damage were found in 8, and bending of the bone was present in 2; the boundary of the focus was clear in 1. According to the MRI examination, the lesions were larger than 5 cm (61.54%),
Conventional imaging features of desmoid tumors

Desmoid tumors of the extremities have certain imaging features on CT and MRI. The two imaging techniques can be combined to improve the diagnostic accuracy, achieve a comprehensive diagnosis of the disease in the clinical practice, and reduce the risk of missed diagnosis or misdiagnosis. In addition, their use can ensure timely diagnosis and treatment.

**Key Words:** Soft tissue desmoid tumor of the extremities; Clinical features; Imaging examination; Computed tomography; Magnetic resonance imaging

**CONCLUSION**

Desmoid tumors of the extremities have certain imaging features on CT and MRI. The two imaging techniques can be combined to improve the diagnostic accuracy, achieve a comprehensive diagnosis of the disease in the clinical practice, and reduce the risk of missed diagnosis or misdiagnosis. In addition, their use can ensure timely diagnosis and treatment.

**INTRODUCTION**

Desmoid fibroma is a rare soft tissue tumor originating from the aponeurosis, fascia, and muscle, and it is also known as aponeurotic fibroma, invasive fibroma, or ligamentous fibroma. The nature of desmoid tumors is between that of benign and malignant tumors, and distant metastasis is rare. However, these tumors are easily misdiagnosed or the diagnosis is missed because they are locally invasive, they recur frequently after surgery, and their imaging features are similar to those of inflammatory lesions or malignant soft tissue tumors; thus, it is difficult to obtain early diagnosis and treatment[1-3].

The soft tissue of the extremities is an important location of desmoid fibroma. The pathogenesis of desmoid fibroma has not yet been elucidated, and it is considered to be closely related to many factors, such as heredity, hormones, previous surgery, and trauma. Because of its complex imaging features, differential diagnosis is difficult, resulting in a low incidence of preoperative diagnosis[4-6]. Therefore, it is of great significance to clarify the clinical and imaging features of desmoid tumors of the extremities.

This study aimed to analyze the data of patients with desmoid fibroma of the extremities from our hospital in order to clarify the clinical and imaging of this disease.
MATERIALS AND METHODS

General data
We selected 13 patients with desmoid fibroma of the extremities admitted to our hospital between October 2016 and March 2021, including 6 men and 7 women, aged from 19 to 44 years (mean, 31.56 ± 10.97 years). Eight of the cases were primary tumors and five were postoperative recurrences.

Methods
All patients underwent computed tomography (CT), magnetic resonance imaging (MRI), and pathological examination of the lesion.

CT: Both plain and contrast-enhanced CT scans were performed using a Brilliance TM16 (Phillips Medical Systems, Best, The Netherlands) or Toshiba Aquillion TM64 (Toshiba Medical Systems, Otawara, Japan) helical CT system with the following settings: Pitch 1; layer thickness, 5 mm; matrix, 512 × 512; tube current, 150–250 mA; tube voltage, 120 kV; intravenous injection of 2 mL/kg non-ionic contrast agent (iohexol), at an injection rate of 2–3 mL/s.

MRI: The GE1.5T (Signa, GE Healthcare, United States) or SIEMENS 3T (Siemens Medical Systems, Erlangen, Germany) scanner with an acquisition matrix of 320 × 224 or 256 × 256 were used to perform the MRI scans. Different coils and scanning fields were taken according to the location or size of the focus. Conventional scanning used a spin echo/fast spin echo sequence to obtain coronal/sagittal, and cross-sectional T1- and T2-weighted images (T1WI and T2WI); in specific conditions, a fat-suppression sequence was also used. The parameters for the coronal/sagittal and cross-sectional T1WI were: Echo time (TE), 9.2/2.2 ms; repetition time (TR), 207/504 ms. For the coronal/sagittal and cross-sectional T2WI, the parameters were: TE, 68.9/80 ms; TR, 3340/3232 ms. The gradient-echo-T1WI and short tau inversion recovery sequences were routinely used for enhanced scanning, including in the coronal/sagittal and cross-sectional planes. Dynamic enhanced LAVA or VIBA sequences were performed for abdominal lesions, and 0.2 mL/kg contrast agent was administered intravenously through the forearm using a high-pressure syringe at a rate of 2.0 mL/s.

Pathological examination: All patients underwent surgical resection, and the removed lesions were fixed with a 4% formaldehyde solution, paraffin-embedded, and stained with hematoxylin and eosin and for immunohistochemical markers. In addition, the pathological sections were evaluated repeatedly by the same pathologist with rich clinical experience.

Data collection
We collected and statistically analyzed the data on the (1) diameter and distribution of the lesions; (2) morphology of the lesion and its relationship with surrounding structures; (3) MRI and CT findings of desmoid tumors of the extremities; and (4) pathological features of the lesions.

Statistical analysis
The data were analyzed using IBM SPSS Statistics for Windows (version 22.0. Armonk, NY: IBM Corp.). Measurement data are expressed as the mean ± SD, and were analyzed using the t-test; counting data are expressed as n (%), and were analyzed using the χ² test. A P value < 0.05 was considered statistically significant.

RESULTS

Diameter and distribution of lesions
Among the 13 patients with desmoid tumors of the extremities, the lesion diameter ranged from 1.7 to 8.9 cm, with an average of 5.35 ± 2.39 cm. All lesions were located in the deep muscular space, with a relatively high frequency in the left and right forearms, each accounting for 23.08% of cases. The specific distribution is shown in Table 1.

Shape of the lesion and its relationship with the surrounding structures
The lesion was “patchy” in 1 case, irregular in 10, and quasi-round in 2. The boundary between the lesion and surrounding soft tissue was blurred in 10 cases, with the lesion
Table 1 Distribution of lesions in this group

| Location      | n (%)|
|---------------|-----|
| Left forearm  | 3 (23.08) |
| Right forearm | 3 (23.08) |
| Left hip      | 2 (15.38) |
| Right hip     | 1 (7.69) |
| Right thigh   | 1 (7.69) |
| Left hand     | 1 (7.69) |
| Left foot     | 1 (7.69) |
| Right hand    | 1 (7.69) |
| Total         | 13 (100.00) |

infiltrating along the tissue space and invading the adjacent structures. The edge of the focus showed "beard-like" infiltration in 2 cases, external compression with bone resorption and damage in 8, bending of the bone in 2, and clear boundary in 1.

MRI findings of desmoid tumors of the extremities
The MRI examination revealed that the lesions were larger than 5 cm (61.54%), round or fusiform in shape (84.62%), had an unclear boundary (76.92%), uniform signal (69.23%), inhomogeneous enhancement (84.62%), and "root" or "claw" infiltration (69.23%). Neurovascular bundle invasion occurred in 30.77% of cases (Table 2).

CT findings of desmoid tumors of the extremities
The CT examination showed that the lesions had a slightly lower density (69.23%), higher degree of enhancement (61.54%), and unclear boundary (84.62%); a CT value < 50 Hu was encountered in 53.85% of lesions, and the enhancement was uneven in 53.85% of cases (Table 3).

Pathological examination
All 13 patients underwent surgical resection of the lesion and biopsy. The pathological examination revealed fibroblasts and myofibroblasts arranged in strands and bundles, no obvious atypia, and occasional karyokinesis. Collagen tissue was seen between cells, and there were disparities in the proportion of tumor cells and collagen tissue across different regions. Some areas were rich in collagen tissue and had few tumor cells, similar to the structure of aponeuroses or ligaments, with tumor cells invading the surrounding tissue.

DISCUSSION
Desmoid tumors of the extremities are benign soft tissue tumors that often occur in the deep fascia, aponeurosis, and muscle. This disease is also referred to as invasive fibroma because of its local invasiveness[7,8]. There is a correlation between the morphology of the desmoid fibroma and its growth pattern. Most infiltrative lesions have an irregular morphology and clear boundaries, and grow along the intermuscular space, enclosing blood vessels, nerves, and other adjacent structures[9, 10]. In the current study, the boundary between the lesion and surrounding soft tissue was blurred in 10 cases, with the lesion infiltrating along the tissue space and invading the adjacent structures. The edge of the focus showed "beard-like" infiltration in 2 cases; bone resorption and damage were found in 8, and bending of the bone was present in 2; the boundary of the focus was clear in 1. This finding is consistent with the results of other relevant studies[11,12].

Imaging examination by CT or MRI plays an important role in the diagnosis and treatment of desmoid tumors of the extremities. Image-guided needle biopsy is commonly used for desmoid tumors and it can provide an objective basis for the diagnosis and treatment of the disease. However, it is an invasive diagnostic and therapeutic procedure that may cause complications. It is not suitable for elderly or gravely ill patients. Furthermore, its clinical application is limited in more severe cases.
Table 2 Magnetic resonance imaging findings of soft tissue desmoid tumors of the extremities

| Manifestation                                      | n (%)     |
|----------------------------------------------------|-----------|
| Diameter                                           |           |
| > 5 cm                                             | 8 (61.54) |
| < 5 cm                                             | 5 (38.46) |
| Shape                                              |           |
| Quasi-circular or shuttle-shaped                    | 11 (84.62)|
| Irregular shape                                     | 2 (15.38) |
| Boundary                                           |           |
| Clear                                              | 3 (23.08) |
| Unclear                                            | 10 (76.92)|
| Signal                                             |           |
| Uneven                                             | 4 (30.77) |
| Uniform                                            | 9 (69.23) |
| Enhancement mode                                   |           |
| Uneven                                             | 11 (84.62)|
| Uniform                                            | 2 (15.38) |
| "Root" or "claw" infiltration                      | 9 (69.23) |
| Invasion of neurovascular bundle                   | 4 (30.77) |

MRI: Magnetic resonance imaging.

Although CT and MRI are commonly used in desmoid tumors of the extremities, some researchers have shown that CT scans of desmoid tumors reveal no typical features; however, the CT scan findings show a certain correlation with the histological composition of the lesions, namely, their degree of enhancement is slightly higher than or equal to that of muscle tissue, they show various types of enhancement after contrast administration and soft tissue density, and on plain scans they appear iso-, hypo-, or hyper-dense relative to the density of the surrounding muscles. In addition, the CT scan shows inhomogeneous or homogenous tissue and the focus lacks a clear boundary; the focus generally shows an infiltrative growth pattern into the surrounding muscles[13-15]. MRI has a higher soft tissue resolution, therefore it can effectively detect the focus and its relationship with the surrounding structures, and provide an objective basis for the differential diagnosis and treatment choice, as well as for determining the scope of surgical resection and postoperative follow-up[16,17]. Some studies have shown that the CT features of desmoid fibroma are insufficient for diagnosis due to the different tissue components within the lesions; MRI findings are diverse and complex, showing that most tumors have unclear boundaries and a tendency to invade blood vessels, nerves, and bones. These imaging features are similar to those of malignant tumors. Consequently, the disease is easily misdiagnosed as soft tissue sarcoma[18]. Sanchez-Mete et al[19] pointed out that soft tissue sarcoma grows rapidly, insufficient blood supply inside the tumor can cause necrosis, and the focus shows expansive growth, compressing the surrounding tissue. Therefore, sarcoma has a clear boundary, while osteofibroma has less necrosis and the invasive growth results in the lack of a clear boundary between the tumor and surrounding tissue; additionally, a band of low signal intensity on T1WI and T2WI can be seen. Thus, MRI can be used for the differential diagnosis between osteofibroma and soft tissue sarcoma.

In the current study, it was found that the CT features of desmoid tumors of the extremities exhibited a slightly low density, mild enhancement, unclear boundary, and uneven enhancement after contrast administration. Their imaging features on MRI were round or fusiform shape, unclear boundary, uniform signal, uneven enhancement, "tree root" or "claw" infiltration, and invasion of the neurovascular bundles. Sehgal et al[20] confirmed that desmoid tumors of the extremities should be differentiated from benign soft tissue tumors, such as neurogenic tumors and
Table 3 Computed tomography findings of soft tissue desmoid tumors of the extremities

| Manifestation          | n (%) |
|------------------------|-------|
| Focus density          |       |
| Slightly lower density | 9 (69.23) |
| Equal density          | 2 (15.38) |
| Slightly higher density| 2 (15.38) |
| Strengthening degree   |       |
| Mild strengthening     | 8 (61.54) |
| Moderate strengthening | 3 (23.08) |
| Obvious strengthening  | 2 (15.38) |
| Boundary               |       |
| Unclear                | 11 (84.62) |
| Clear                  | 2 (15.68) |
| CT                     |       |
| < 50 Hu                | 7 (53.85) |
| ≥ 50 Hu                | 6 (46.15) |
| Enhancement mode       |       |
| Uneven                 | 7 (53.85) |
| Uniform                | 6 (46.15) |

CT: Computed tomography.

hemangiomas, and malignant tumors, such as synovial sarcoma, rhabdomyosarcoma, lymphoma, and fibrous histiocytoma. It has been shown through MRI examinations that the above tumors have specific manifestations, such as the tortuous flow void signal in hemangioma, and significant enhancement after contrast administration. On the other hand, most malignant soft tissue tumors show inhomogeneous long T1 and long T2 signals, T2 signal intensity higher than that of fat, and calcification or cystic necrosis. These features can be used to diagnose desmoid tumors of the extremities. Therefore, CT scans and MR images can show certain imaging features of desmoid tumors of the extremities, but MRI can be used to identify the lesions more effectively and provide an objective basis for the diagnosis. Additionally, MRI has a higher soft tissue resolution and application value for soft tissue tumors such as desmoid fibroma (especially in the extremities or head and neck). It is also suitable for younger patients, in whom the use of ionizing radiation should be avoided or who are allergic to iodine contrast agents. However, CT and MRI can be combined to maximize the diagnostic accuracy and sensitivity of the differential diagnosis, as well as to reduce the incidence of missed diagnosis or misdiagnosis.

CONCLUSION

Generally, desmoid tumors of the extremities show certain imaging features on CT and MRI examination. These two imaging techniques can be combined to improve diagnostic accuracy, achieve a comprehensive diagnosis of the disease, reduce the risk of missed diagnosis or misdiagnosis, and ensure timely diagnosis and treatment. This study had certain limitations, such as its single-center nature and the small sample size. Therefore, further investigations on larger samples are needed to confirm whether the results of our study are applicable on a broader scale.
ARTICLE HIGHLIGHTS

Research background
Desmoid fibroma is a rare soft tissue tumor originating from the aponeurosis, fascia, and muscle, and it is also known as aponeurotic fibroma, invasive fibroma, or ligamentous fibroma.

Research motivation
The soft tissue of the extremities is an important location of desmoid fibroma; its pathogenesis has not yet been elucidated.

Research objectives
This study aimed to analyze the data of patients with desmoid fibroma of the extremities from our hospital in order to clarify the clinical and imaging of this disease.

Research methods
We selected 13 patients with desmoid fibroma of the extremities. All patients underwent computed tomography (CT), magnetic resonance imaging (MRI), and pathological examination of the lesion.

Research results
Neurovascular tract invasion was present in 30.77% of cases. CT examination showed that the desmoid tumors had a slightly lower density, higher enhancement, and unclear boundary; a CT value < 50 Hu was present in 53.85% of lesions, and the enhancement was uneven in 53.85% of cases. Microscopically, fibroblasts and myofibroblasts were arranged in strands and bundles, without obvious atypia but with occasional karyotyping; cells were surrounded by collagen tissue.

Research conclusions
Desmoid tumors of the extremities have certain imaging features on CT and MRI. The two imaging techniques can be combined to improve the diagnostic accuracy, achieve a comprehensive diagnosis of the disease in the clinical practice, and reduce the risk of missed diagnosis or misdiagnosis.

Research perspectives
Further investigations on larger samples are needed to confirm whether the results of our study are applicable on a broader scale.

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