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REVIEW

- 8627 Time to give up traditional methods for the management of gastrointestinal neuroendocrine tumours
Yozgat A, Kekilli M, Altay M

MINIREVIEWS

- 8647 Healthcare practice strategies for integrating personalized medicine: Management of COVID-19
Liu WY, Chien CW, Tung TH
- 8658 Clinical application of repetitive transcranial magnetic stimulation for post-traumatic stress disorder: A literature review
Cheng P, Zhou Y, Xu LZ, Chen YF, Hu RL, Zou YL, Li ZX, Zhang L, Shun Q, Yu X, Li LJ, Li WH
- 8666 Pros and cons of continuous glucose monitoring in the intensive care unit
Sun MT, Li IC, Lin WS, Lin GM

ORIGINAL ARTICLE**Clinical and Translational Research**

- 8671 Prognostic implications of ferroptosis-associated gene signature in colon adenocarcinoma
Miao YD, Kou ZY, Wang JT, Mi DH

Retrospective Study

- 8694 Cefoperazone sodium/sulbactam sodium *vs* piperacillin sodium/tazobactam sodium for treatment of respiratory tract infection in elderly patients
Wang XX, Ma CT, Jiang YX, Ge YJ, Liu FY, Xu WG
- 8702 Modified Gant procedure for treatment of internal rectal prolapse in elderly women
Xu PP, Su YH, Zhang Y, Lu T
- 8710 Clinical and imaging features of desmoid tumors of the extremities
Shi Z, Zhao XM, Jiang JM, Li M, Xie LZ
- 8718 Retrospective analysis of surgically treated pT4b gastric cancer with pancreatic head invasion
Jin P, Liu H, Ma FH, Ma S, Li Y, Xiong JP, Kang WZ, Hu HT, Tian YT
- 8729 Development of a random forest model for hypotension prediction after anesthesia induction for cardiac surgery
Li XF, Huang YZ, Tang JY, Li RC, Wang XQ

Clinical Trials Study

- 8740** Effects of mindful breathing combined with sleep-inducing exercises in patients with insomnia
Su H, Xiao L, Ren Y, Xie H, Sun XH

Observational Study

- 8749** Chronic hepatitis-C infection in COVID-19 patients is associated with in-hospital mortality
Ronderos D, Omar AMS, Abbas H, Makker J, Baiomi A, Sun H, Mantri N, Choi Y, Fortuzi K, Shin D, Patel H, Chilimuri S
- 8763** Midazolam dose is associated with recurrence of paradoxical reactions during endoscopy
Jin EH, Song JH, Lee J, Bae JH, Chung SJ

CASE REPORT

- 8773** Isolated mass-forming IgG4-related sclerosing cholangitis masquerading as extrahepatic cholangiocarcinoma: A case report
Song S, Jo S
- 8782** *Samonella typhi* infection-related appendicitis: A case report
Zheng BH, Hao WM, Lin HC, Shang GG, Liu H, Ni XJ
- 8789** ACTA2 mutation is responsible for multisystemic smooth muscle dysfunction syndrome with seizures: A case report and review of literature
Yang WX, Zhang HH, Hu JN, Zhao L, Li YY, Shao XL
- 8797** Whole-genome amplification/preimplantation genetic testing for propionic acidemia of successful pregnancy in an obligate carrier Mexican couple: A case report
Neumann A, Alcantara-Ortigoza MA, González-del Angel A, Zarate Díaz NA, Santana JS, Porchia LM, López-Bayghen E
- 8804** Is mannitol combined with furosemide a new treatment for refractory lymphedema? A case report
Kim HS, Lee JY, Jung JW, Lee KH, Kim MJ, Park SB
- 8812** Successful treatment of floating splenic volvulus: Two case reports and a literature review
Sun C, Li SL
- 8820** Removal of "ruptured" pulmonary artery infusion port catheter by pigtail catheter combined with gooseneck trap: A case report
Chen GQ, Wu Y, Zhao KF, Shi RS
- 8825** Isolated neutropenia caused by copper deficiency due to jejunal feeding and excessive zinc intake: A case report
Ohmori H, Kodama H, Takemoto M, Yamasaki M, Matsumoto T, Kumode M, Miyachi T, Sumimoto R
- 8831** Diagnosis and treatment of eosinophilic fasciitis: Report of two cases
Song Y, Zhang N, Yu Y
- 8839** Familial left cervical neurofibromatosis 1 with scoliosis: A case report
Mu X, Zhang HY, Shen YH, Yang HY

- 8846** Successful treatment after toxic epidermal necrolysis induced by AZD-9291 in a patient with non-small cell lung cancer: A case report
Li W, He X, Liu H, Zhu J, Zhang HM
- 8852** Anesthesia management in a pediatric patient with Becker muscular dystrophy undergoing laparoscopic surgery: A case report
Peng L, Wei W
- 8858** Diagnosis of upper gastrointestinal perforation complicated with fistula formation and subphrenic abscess by contrast-enhanced ultrasound: A case report
Qiu TT, Fu R, Luo Y, Ling WW
- 8864** Adenomyoepithelioma of the breast with malignant transformation and repeated local recurrence: A case report
Oda G, Nakagawa T, Mori M, Fujioka T, Onishi I
- 8871** Primary intracranial synovial sarcoma with hemorrhage: A case report
Wang YY, Li ML, Zhang ZY, Ding JW, Xiao LF, Li WC, Wang L, Sun T
- 8879** Lumbar infection caused by *Mycobacterium paragordoniae*: A case report
Tan YZ, Yuan T, Tan L, Tian YQ, Long YZ
- 8888** Primary intratracheal neurilemmoma in a 10-year-old girl: A case report
Wu L, Sha MC, Wu XL, Bi J, Chen ZM, Wang YS
- 8894** Ovarian pregnancy rupture following ovulation induction and intrauterine insemination: A case report
Wu B, Li K, Chen XF, Zhang J, Wang J, Xiang Y, Zhou HG
- 8901** Delayed diagnosis of imperforate hymen with huge hematocolpometra: A case report
Jang E, So KA, Kim B, Lee AJ, Kim NR, Yang EJ, Shim SH, Lee SJ, Kim TJ
- 8906** Acute pancreatitis with hypercalcemia caused by primary hyperparathyroidism associated with paraneoplastic syndrome: A case report and review of literature
Yang L, Lin Y, Zhang XQ, Liu B, Wang JY
- 8915** Use of a modified tracheal tube in a child with traumatic bronchial rupture: A case report and review of literature
Fan QM, Yang WG
- 8923** Isolated liver metastasis detected 11 years after the curative resection of rectal cancer: A case report
Yonenaga Y, Yokoyama S
- 8932** Severe bleeding after operation of preauricular fistula: A case report
Tian CH, Chen XJ
- 8938** Secondary aorto-esophageal fistula initially presented with empyema after thoracic aortic stent grafting: A case report
Wang DQ, Liu M, Fan WJ

- 8946** Disruption of sensation-dependent bladder emptying due to bladder overdistension in a complete spinal cord injury: A case report

Yoon JY, Kim DS, Kim GW, Won YH, Park SH, Ko MH, Seo JH

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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 case, 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%),

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round or fusiform in shape (84.62%), had an unclear boundary (76.92%), showed uniform signal (69.23%), inhomogeneous enhancement (84.62%), and "root" or "claw" infiltration (69.23%). Neurovascular tract invasion was present in 30.77% of cases. CT examination showed that the desmoid tumors had slightly a lower density (69.23%), higher enhancement (61.54%), and unclear boundary (84.62%); 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. There were disparities in the proportion of collagen tissue in different regions, with abundant collagen tissue and few tumor cells in some areas, similar to the structure of aponeuroses or ligaments, and tumor cells invading the surrounding tissues.

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.

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

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Core Tip: Generally, soft tissue desmoid tumors of the extremities show certain imaging features on computed tomography and magnetic resonance imaging. These two imaging techniques can be combined to comprehensively diagnose the disease, improve the diagnostic accuracy, reduce the risk of missed diagnosis or misdiagnosis, and ensure that patients are diagnosed and treated as soon as possible.

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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 \pm SD, and were analyzed using the *t*-test; counting data are expressed as *n* (%), and were analyzed using the χ^2 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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