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Review

Current Update on the Diagnosis, Management and Pathogenesis of Elastofibroma Dorsi

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Abstract. *Elastofibroma dorsi is an uncommon benign fibroblastic pseudotumor that typically occurs in the subscapular region of middle-aged or older individuals. The pathogenesis is still unclear and a matter of debate. Magnetic resonance imaging can be used as a first-line investigation of the lesion and reveals a lenticular soft-tissue mass with a signal intensity similar to that of skeletal muscle interlaced with strands of fat. Biopsy is not necessary if all pathognomonic criteria are present. A conservative "wait and see" attitude is reasonable and immediate surgery is no more the standard treatment of elastofibroma dorsi. This review provides an updated overview of the diagnosis, management and pathogenesis of elastofibroma dorsi. We also discuss recent advances in our understanding of genomic alterations in elastofibroma dorsi.*

Elastofibroma dorsi is a rare benign soft-tissue pseudotumor first described by Jarvi and Saxen in 1961 (1). It belongs to the fibroblastic/myofibroblastic tumor group according to the 2020 World Health Organization Classification of Tumours of Soft Tissue and Bone (2). The lesion usually arises beneath the rhomboid major and latissimus dorsi muscles adjacent to the inferior angle of the scapula. Current knowledge on the genetic features of this peculiar condition is mostly limited. In this review, we present an updated overview of the diagnosis, management and pathogenesis of elastofibroma dorsi. We also discuss recent advances in our understanding of genomic alterations in elastofibroma dorsi.

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Clinical Features

Elastofibroma dorsi may be an incidental radiologic finding. It has a peak incidence in the seventh to eighth decades of life, with a female predominance. Children are rarely affected. Elastofibroma dorsi typically presents as a firm, slow-growing, painless mass and can occur bilaterally. The process has a strong tendency to develop between the inferomedial portion of the scapula and the chest wall at the level of sixth to eighth ribs (Figure 1). When symptoms are present, they are usually mild pain, discomfort or clunking of the scapula during movement of the shoulder. The diameter ranges from 2 to 15 cm (2). In our experience, spontaneous regression is extremely rare. There is no metastatic potential and no reports of malignant transformation.

Radiologic Findings

Various imaging modalities have been applied for the detection and follow-up of elastofibroma dorsi. In our experience, magnetic resonance imaging (MRI) is the mainstay of imaging in elastofibroma dorsi. It is essential to be familiar with the key imaging findings of elastofibroma dorsi for its accurate diagnosis and appropriate management. Plain radiographs are usually normal but may occasionally show a soft-tissue mass without calcification. Although reported, bone erosion is extremely unusual. Four ultrasound patterns have been detected (3). A heterogenous fasciculated pattern is most frequently seen. Color Doppler evaluation has not shown significant blood flow (3). Computed tomography (CT) usually shows a poorly defined, crescent shaped, heterogeneous soft-tissue mass with attenuation similar to that of skeletal muscle and linear streaks of fat attenuation (Figure 2). In our experience, CT is less sensitive than MRI for visualizing the streaks of adipose tissue. MRI clearly demonstrates a lenticular, poorly circumscribed, heterogeneous soft-tissue mass (4-6). On both T1- and T2-weighted images (Figure 3), the lesion

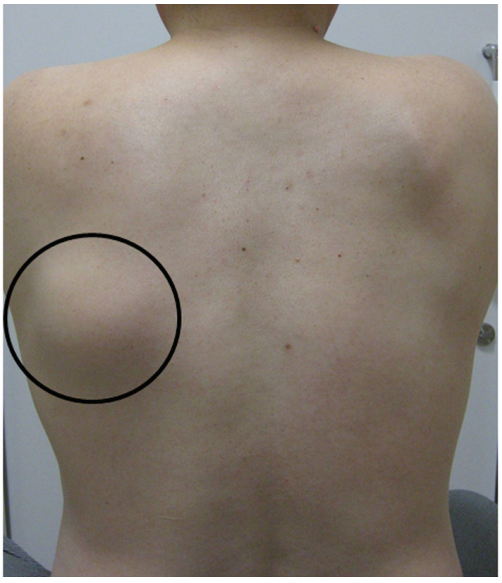

Figure 1. *Clinical photograph reveals a palpable mass (circle).*

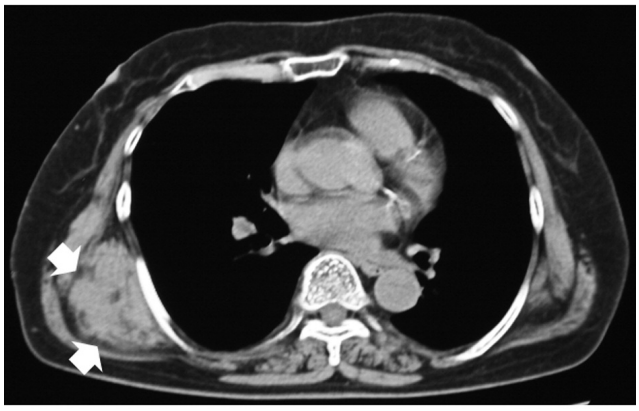

Figure 2. *Axial computed tomography shows a poorly circumscribed, lenticular mass (arrows) with attenuation similar to that of skeletal muscle and linear streaks of fat attenuation.*

displays intermediate signal intensity similar to that of skeletal muscle with interspersed linear and curvilinear areas of high signal intensity. The absence of abnormal diffusion restriction has been reported (6). Gadolinium-enhanced MRI reveals heterogeneity containing areas with and without contrast enhancement. In our opinion, contrast administration is not necessary to characterize the lesion. Integrated positron-emission tomography (PET)/CT images show mild to moderate fluorodeoxyglucose (FDG) uptake by the lesion (7, 8). It is known that elastofibroma dorsi is often incidentally detected as an FDG-avid mass on PET/CT (9).

Histological and Immunohistochemical Characteristics

Elastofibroma dorsi presents a typical macroscopical and histological aspect. Macroscopically, it appears as an ill-defined, rubbery to firm mass. Cut surface displays strands of gray-white fibrous tissue with variable amounts of entrapped yellow adipose tissue (Figure 4). Histologically, the lesion is composed of a mixture of paucicellular fibrocollagenous tissue, a large number of abnormal elastic fibres, scattered spindle cells and entrapped mature fat cells (Figure 5A). Cystic change within the lesion may be seen. Mitotic figures and nuclear pleomorphism are not found. Elastica van Gieson stain reveals numerous globules and branched or unbranched fibres (Figure 5B). Immunohistochemically, the spindle cells are positive for vimentin, CD34 and lysozyme (10). Expression of smooth muscle actin, desmin and S-100 protein is absent.

Cytogenetic and Molecular Genetic Findings

Cytogenetic analyses of elastofibroma dorsi have shown significant chromosomal instability, with clonal and nonclonal structural changes (11-13). Aberrations of the short arm of chromosome 1 are particularly notable. Previously, we identified the presence of DNA copy number changes in 9 of 27 (33%) cases of elastofibroma dorsi (14). The most common recurrent change was a gain of Xq12-q22. In 2006, a nonrandom inactivation of X-chromosome-linked androgen receptor gene was detected in 2 cases of elastofibroma dorsi (10). In 2010, Hernández *et al.* (15) described losses of 1p36-p31, 19p13.3-q13.1 and 22q11-q13 and a gain of 6p25-q25. In that study, deletions of calcium sensing receptor (*CASR*), Glutathione S-transferase Pi 1 (*GSTP1*) and BRCA2 DNA repair associated (*BRCA2*) and gains of APC regulator of WNT signaling pathway (*APC*) and phenylalanine hydroxylase (*PAH*) were also identified. These findings suggest that elastofibroma dorsi is a possible neoplastic process. However, further studies are needed to better understand the correlation between certain genomic alterations and distinct biological behavior.

Diagnosis and Management

In our experience, the lesion can be confidently diagnosed by radiologic examination alone (ideally MRI) if typical clinical features are present. Although we performed biopsy to obtain a diagnosis of elastofibroma dorsi in the past, this is no longer our standard practice. We propose that biopsy is necessary only when the lesion has enlarged rapidly within

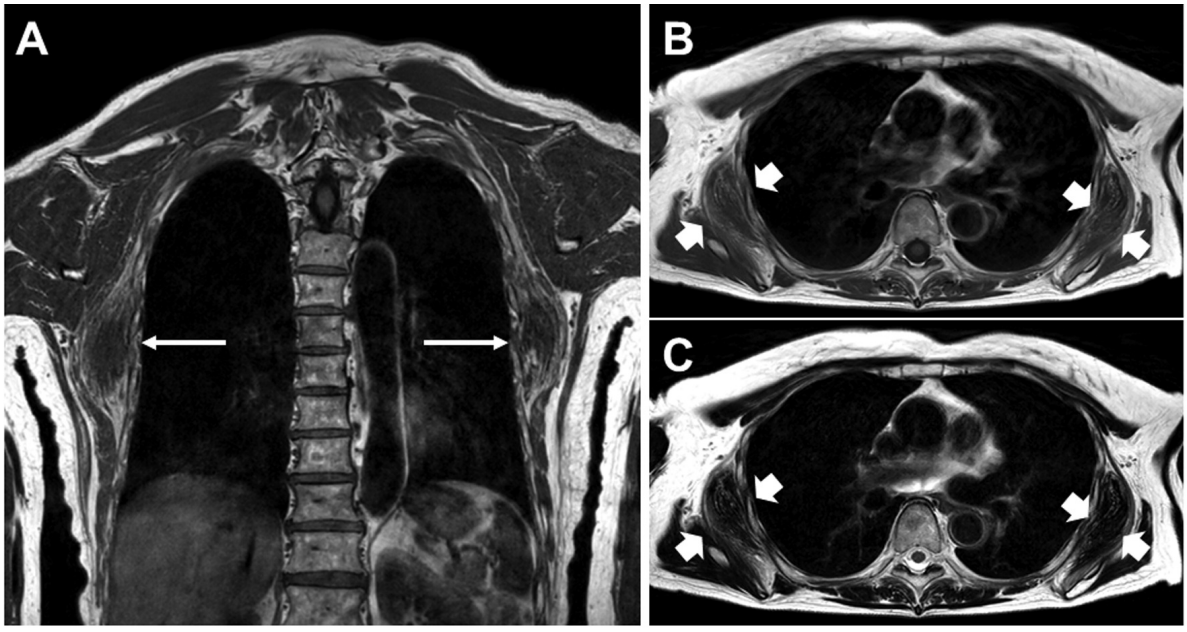

Figure 3. *Magnetic resonance images of elastofibroma dorsi. (A) Coronal T1-weighted image reveals bilateral lesions (arrows) in the subscapular region. Corresponding axial T1-weighted (B) and T2-weighted (C) images show bilateral heterogeneous soft-tissue masses (arrows) with a signal intensity approximately equal to that of skeletal muscle, interlaced with areas of signal intensity similar to that of fat.*

a few months or imaging findings are atypical. Once the diagnosis is established, follow-up radiologic examinations will be unnecessary.

The differential diagnosis for elastofibroma dorsi is limited and includes fibrolipoma and desmoid-type fibromatosis. Fibrolipoma is a rare variant of lipoma and has a prominent fibrous tissue component in addition to mature adipose tissue element (16). If fibrolipoma has a marked hypointense signal on all pulse sequences, it may have imaging findings that overlap with those of elastofibroma dorsi. Desmoid-type fibromatosis is a locally aggressive fibroblastic neoplasm that can occur in the subscapular location (17). In our experience, the presence of linear fascial extensions and low signal-intensity bands on MRI is characteristic of desmoid-type fibromatosis. Histologically, unlike elastofibroma dorsi, these neoplasms lack the abnormal elastic element.

The management of elastofibroma dorsi is somewhat controversial. In our experience, simple observation is a treatment option that can always be considered. It is reasonable to wait at least a year or two in order to understand whether the disease is stable or progressing. On the other hand, Nagano *et al*. (18) proposed that surgery should only be recommended for symptomatic patients. Chandrasekar *et al*. (19) suggested that patient preference is also important. If symptomatic patients choose to have surgery, a marginal excision is sufficient and can be performed with minimal morbidity. However, a high incidence of postoperative complications such as seroma or hematoma has been reported in the literature (18, 20). We recommend the use of postoperative wound drainage and compression bandage to prevent these complications (4).

Pathogenesis

There are various opinions regarding the pathogenesis of elastofibroma dorsi. Familial occurrence has been reported in Japan and Europe (21-24). In the largest series of the lesion, 55 of 170 (32%) cases occurred within the same family lines (21). These findings suggested a genetic predisposition of this lesion. However, many authors believe that elastofibroma dorsi is caused by friction of the lower scapula against the thoracic wall due to repetitive minor trauma and manual labor (25-27). In addition, some other factors may underlie its pathogenesis, including vascular insufficiency, elastotic degeneration of collagen and abnormal elastotic fibrinogenesis (2, 25, 28-30). Recently, Di Vito *et al*. (31) reported that stroma and interspersed spindle cells of elastofibroma dorsi were positive for periostin and tenascin-C. It is possible that these two matricellular proteins may be involved in the development of fibrosis. In that study, the presence of tryptase-positive mast cells was also identified throughout the lesion. However, the precise role of mast cells in elastofibroma dorsi is not yet clarified.

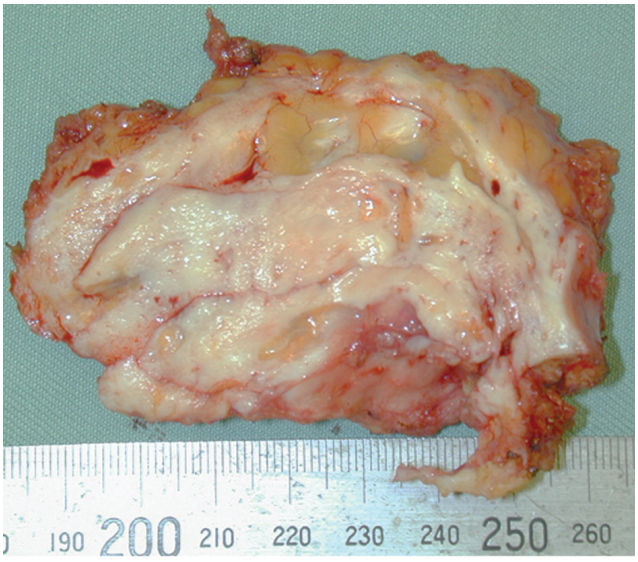

Figure 4. *The cut surface of elastofibroma dorsi demonstrates a variegated appearance with an admixture of gray-white fibrous tissue and intervening yellow fat.*

Conclusion

Elastofibroma dorsi is a distinctive benign fibroblastic pseudotumor and its associated prognosis is excellent with no recurrence. MRI is the mainstay of diagnosis for this lesion. We strongly recommend that a conservative wait and see strategy should be the front-line approach to newly diagnosed patients, irrespective of existing pain or other clinical symptoms. Recurrent abnormalities of 1p and Xq are prominent in elastofibroma dorsi. Further studies are required to determine the biological consequences of these genomic alterations in elastofibroma dorsi.

Conflicts of Interest

The Authors declare no conflicts of interest associated with this article.

Authors' Contributions

JN researched the literature and drafted the article. SN revised the article. KN performed the histological evaluations and reviewed the article. TY reviewed the article. All Authors read and approved the final article.

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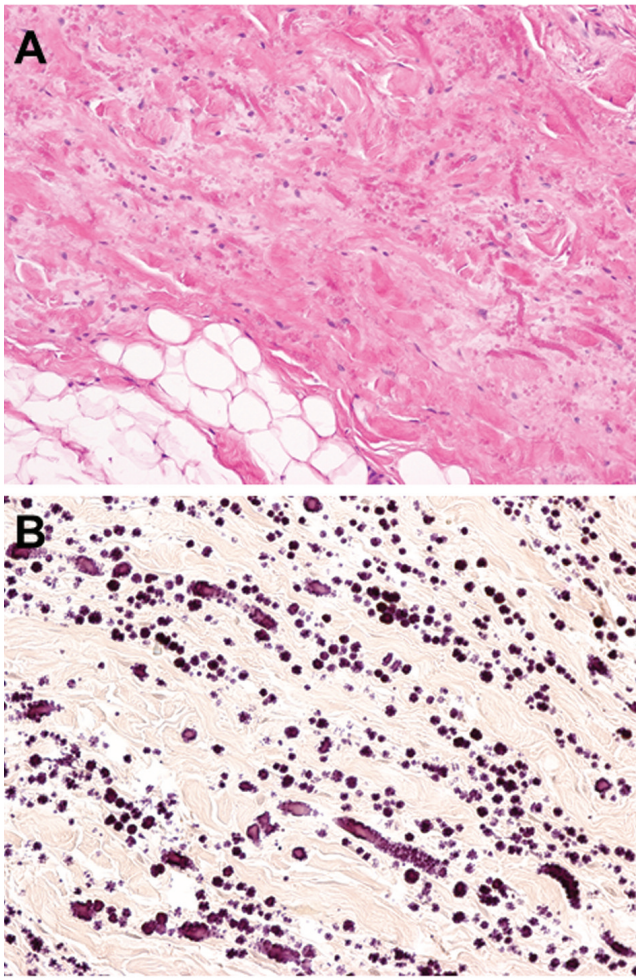

Figure 5. *Histological findings of elastofibroma dorsi. (A) The lesion is composed of fibrocollagenous tissue containing eosinophilic elastic fibres and scattered fibroblasts with a variable amount of mature adipose tissue (hematoxylin and eosin stain, original magnification ×100). (B) Elastica van Gieson stain displays numerous globules and abnormal elastic fibres (original magnification ×200).*

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