Bilateral Familial Elastofibroma Dorsi: Is Genetic Abnormality Essential?
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Elastofibroma dorsi is a rare, slowly growing, benign soft tissue tumor located in the periscapular region on the chest wall. Although it is generally unilateral, it may be bilateral in 10% of cases. The tumor is located between the costa and the chest wall muscles. The patients generally describe back and shoulder pain that increases with movement. Three sisters, aged 58, 61, and 63 years, who were admitted to our clinic with of bilateral swelling and pain in the infrascapular region, received diagnoses of elastofibroma dorsi after radiologic examination. Bilateral elastofibroma dorsi resection was performed on the three sisters. Inasmuch as cases of elastofibroma dorsi demonstrate familial features, a genetic examination was conducted; however, prominent chromosomal instability was not detected. It is emphasized that the disease might be familial; however, chromosomal changes may not be present.

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Elastofibroma dorsi (ED) was first defined by Jarvi and Saxen [1] in 1961 as a slowly growing, uncapsulated, benign, and nonrecurring soft tissue tumor after local surgical resection. Although the incidence in the autopsy series is reported at different rates, the true incidence could not be precisely revealed. ED, which is generally seen in women in their fourth to sixth decades, is characterized by the proliferation of the elastin component of fibrous tissue.

Although the lesion is usually located in the infrascapular region, it may also be seen in the olecranon, deltoid muscle, lateral chest wall, breast, foot, and some visceral organs [2–5]. Bilateralism occurs at a rate of 10% [2, 5].

Three sisters, aged 58, 61, and 63 years, who were admitted with back pain and bilateral subscapular mass, were operated on with the preliminary diagnosis of ED after radiologic examination. Chromosomal abnormality was investigated in the sisters for genetic aspects. They were admitted to our polyclinic 6 months apart with bilateral subscapular palpable mass and back pain. Upon physical examination, soft and mobile mass lesions of the largest size, ranging between 4 cm and 10 cm, were detected in the bilateral subscapular regions. It was learned from their histories that they were among nine siblings, eight of whom were female and one male; no similar pathologic condition in the other six siblings. Each patient lived in the city center, and their sociocultural level was intermediate to high. One of the patients was a teacher and had lived in a city for a long time apart from her siblings, who were housewives. Although none of them were reported to have a job or a hobby that needed serious physical effort to perform, frequent housework was thought to be the factor for microtrauma.

Blood biochemistry values and respiratory function test results were normal. Upon computed tomography (CT) (Fig 1) and magnetic resonance imaging (MRI) of the chest, uncapsulated soft tissue masses, with sizes ranging between 4.3 × 3.2 cm and 10.0 × 4.5 cm, largely isodense with muscle tissue and smooth margins, and in which partial linear fat intensities were observed, were detected in the infrascapular region in the sisters, and the findings were found to be consistent with ED.

In all cases, ED resection was performed simultaneously with bilateral subscapular incision by two chest surgeons in the same session. The lesions were completely removed with penetrative and blunt dissection (Fig 2). The Hemovac drainage tubes that were
inserted in the surgical regions, one each, were removed on the second postoperative day. ED was diagnosed by the fibrous tissue intertwined with fatty tissue upon histopathologic examination.

For genetic analysis, examinations were performed on the preoperative blood samples and postoperative ED materials. The patients were informed about this subject in detail, and informed consents were obtained. Upon receipt of the specimens, the tissues were disaggregated with scalpels and collagenase. The specimens were cultured in RPMI-1640 medium supplemented with 20% fetal bovine serum and antibiotics for 4 to 6 days. Metaphase chromosomes were banded with Giemsa trypsin, and karyotypes were described according to established international guidelines. No complications developed in any of the patients; the 1 patient who was operated on last is in her tenth postoperative month at this writing. The other asymptomatic six siblings were informed of the terms of ED.

Comment

Elastofibroma dorsi is a soft tissue tumor that is mostly seen unilaterally and generally in women over 55 years of age [2, 6]. It is not known why it is more commonly seen in women. The three sisters described here, all of whom were over 55 years of age with bilateral lesions, suggested the possibility of a genetic abnormality.

Although many opinions are proposed in the pathogenesis of ED, there is no consensus about the exact etiologic factor. The most commonly encountered hypothesis is recurrent minor trauma in the subscapular region, which develop by rubbing of the lower end of the scapula on the posterior chest wall. However, ED may also develop in individuals who are not exposed to such trauma. In such cases, reactive fibromatosis, degeneration due to vascular insufficiency, or an enzyme defect may also be suggested. Occasionally, in some cases, proven genetic instability is demonstrated, so it is thought that familial predisposition may be present. As a result, the etiology of ED demonstrates a multifactorial picture [2, 4, 7].

In diagnosis, CT, MR, and ultrasonography may be used. It is emphasized that the most appropriate and distinctive investigation method is MRI (2). On CT, the lesions are observed as heterogeneous soft tissue mass with a density similar to that of muscle tissue, making it difficult to differentiate the surrounding muscle planes from the tumor with distinct margins. The appearance of ED in MRI is as a soft tissue mass with a density similar to that of muscle tissue; however, it consists of linear opacities that belong to the fat tissue. In T1- and T2-weighted images, the appearance of a soft tissue mass consisting of smooth contorted linings of high or intermediate density is characteristic. Preoperative radiologic examinations remove the need for biopsy [4].

Lipoma, hemangioma, metastatic or primary sarcoma, desmoid tumor, neurofibroma, cicatricial fibroma, and fibrous histiocytoma should be considered in the differential diagnosis. Although these initial diagnoses can be considered, excisional biopsy should be the preferred method. In the literature, we are aware of only one case in which recurrence was reported after excision. Complete surgical resection is sufficient in both the definitive diagnosis and the treatment of ED, which is known not to change to malignancy and to metastasize [2].

Very few studies have reported that ED demonstrates familial features [7]. Furthermore, no case report was found that demonstrated both bilateralism and familial features and in which a genetic examination was conducted. This study demonstrates that there may be chromosomal abnormality in familial ED cases that show bilateralism.

Cytogenetic analysis has been performed very rarely in elastofibroma because it is a rarely seen pseudotumor composed of excess collagen and abnormal elastic fibrils. The prominent cytogenetic feature of elastofibroma is remarkably high karyotypic instability, responsible for structural changes involving virtually all chromosomes. Changes are usually random, and when clonal they have always been observed in no more than two cells. The most frequently nonrandomly affected breakpoint is at the 1p32 band, but breakpoints at 1p36 and 3q21 have been recorded [3]. This situation, which represents chromosomal instability, is generally observed at later ages. As a result of cytogenetic examination of the three cases described here, it was found that apparent chromosomal instability had not yet begun in any of them. However, it is possible that such instability may develop in these patients later. Therefore, the researchers believe that it would be useful to follow up on these cases with high-resolution genetic studies. Additionally, there is a need for studies involving more patients to use the cytogenetic modifications as an effective determinant.

References