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CASE REPORT

Chest wall secondary chondrosarcoma caused by malignant degeneration of an enchondroma: case report and literature review

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Introduction: Enchondromas are benign tumors originating in the cartilaginous tissue of the hyaline gristle, rarely located in the chest wall. They sometimes undergo a sarcomatous transformation, becoming secondary chondrosarcomas. **Case presentation**: We present the case of a 53-year-old patient who, following a chest computed tomography scan performed after a thoracic trauma, was diagnosed with an osteolytic tumor at the chondrocostal junction of rib 4. Surgery was performed, with partial straight resection of ribs 3–5. Histopathological examination of the resection piece identified the existence of a chest wall chondrosarcoma on the background of malignant degeneration of an enchondromatosis lesion. The postoperative evolution was favorable, and the patient was discharged on the eighth postoperative day. **Conclusion**: In patients with even asymptomatic chest wall enchondromas, periodic clinical evaluation of these lesions is required, given their risk of malignant degeneration.

Keywords: enchondromatosis, secondary chondrosarcoma, chest wall

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Introduction

Enchondromas are benign tumors that form in the hyaline cartilage, most commonly in the bones of the limbs, and are often asymptomatic [1]. They usually represent single lesions, but there may be multiple enchondromas, as in this case, where the disease is called enchondromatosis. Often, patients with enchondromatosis have deformities of the affected bones and thus the anatomical lesion. The most common are acquired cartilaginous lesions without hereditary substrate, but cases have been cited in the literature with hereditary, autosomal recessive or dominant transmission [2]. This condition is most commonly diagnosed in children, and rarely in adults, with a prevalence in the general population of 1 case per 100,000 people [3].

The most common presentation is asymptomatic, with the lesions discovered by chance when performing x-rays for other conditions. The main risk of these is their malignant transformation. Enchondromas usually turn malignant, causing chondrosarcomas. Their risk of malignant degeneration is estimated in the literature to be 4%, but in the case of multiple enchondromas, it can reach up to 40% [4–6].

The purpose of this article is to present the case of a 53-year-old patient who presented with a case of secondary thoracic wall chondrosarcoma, on the background of malignant degeneration of thoracic wall enchondromatosis lesions.

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Case presentation

A male patient, aged 53, sustained a thoracic trauma by falling, which was the reason for his presentation to the emergency service. Chest x-ray showed an opacity of around 3 cm in diameter at the anterior extremity of the right rib 4. Chest computed tomography (CT) examination revealed an osteolytic lesion at the chondrocostal junction of the right rib 4, which causes the destruction of the bone cortex, without contrast, and invades the parietal pleura. The lesion had a diameter of around 4 cm, and no other lesions were found in the thoraco-abdominal viscera (Figure 1).

The patient had no previously known thoracic wall disorders, and the thoracic clinical examination did not find the existence of lesions that cause deformation of the region. He had been completely asymptomatic before the thoracic trauma.

Due to the osteolytic lesion at the level of rib 4, the therapeutic indication was surgery to remove the costal lesion. After adequate preoperative preparation, surgery was performed, resecting ribs 3–5 within the limits of surgical safety, approximately 2 cm from the edge of the tumor formation. Reconstruction of the chest wall was performed with polypropylene mesh. The postoperative evolution was favorable, with the patient discharged on the eighth postoperative day.

Histopathological examination of the resection piece revealed enchondroma lesions in ribs 3–5, identified by the existence of intracartilaginous and intramedullary nodules, surrounded by a thin blade of bone tissue (Figure 2A). Additionally, at the level of rib 4, areas of secondary chondrosarcoma were identified, the tumor lobes penetrating

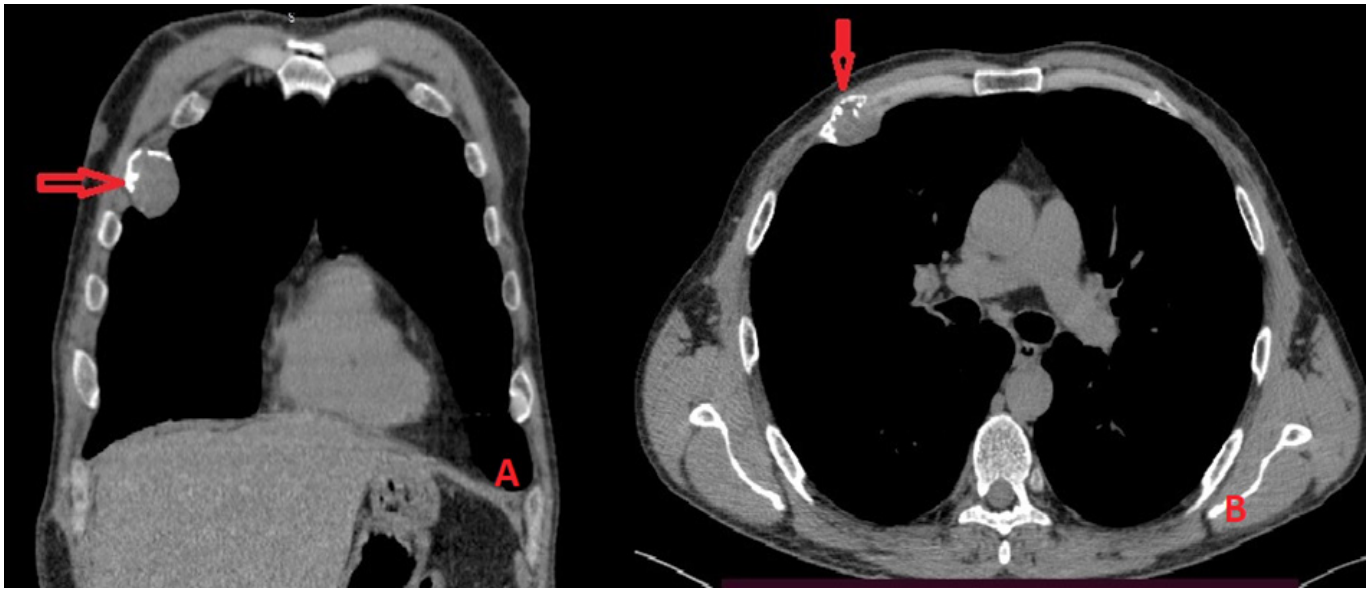

Fig. 1 Chest CT examination: A - coronal section, B - axial section

and infiltrating the pre-existing bone trabeculae (Figure 2B) and tumor infiltrating the periosteum (Figure 2C). At the level of the tumor formation, the chondrocytes were arranged alone or in small groups, showing obvious cyto-nuclear atypia (Figure 2D).

The patient underwent six weeks of adjuvant radiotherapy. The CT simulation was performed on a helical CT (Siemens Somatom Emotion 16) with a 5-mm slice thickness. The target volume included the postoperative bed plus 2-cm expansion in all directions (excluding the lung) and the scar, which received a dose of 50 Gy. In 25 fractions, with a sequential boost up to 60 Gy. On the tumoral bed.

Treatment was delivered by an Elekta Synergy machine, with 6-MV photons, using a dynamic intensity-modulated radiation therapy (IMRT) technique with five fields. To ensure the acuity of treatment delivery, cone-beam computed tomography (CBCT) was performed daily in the

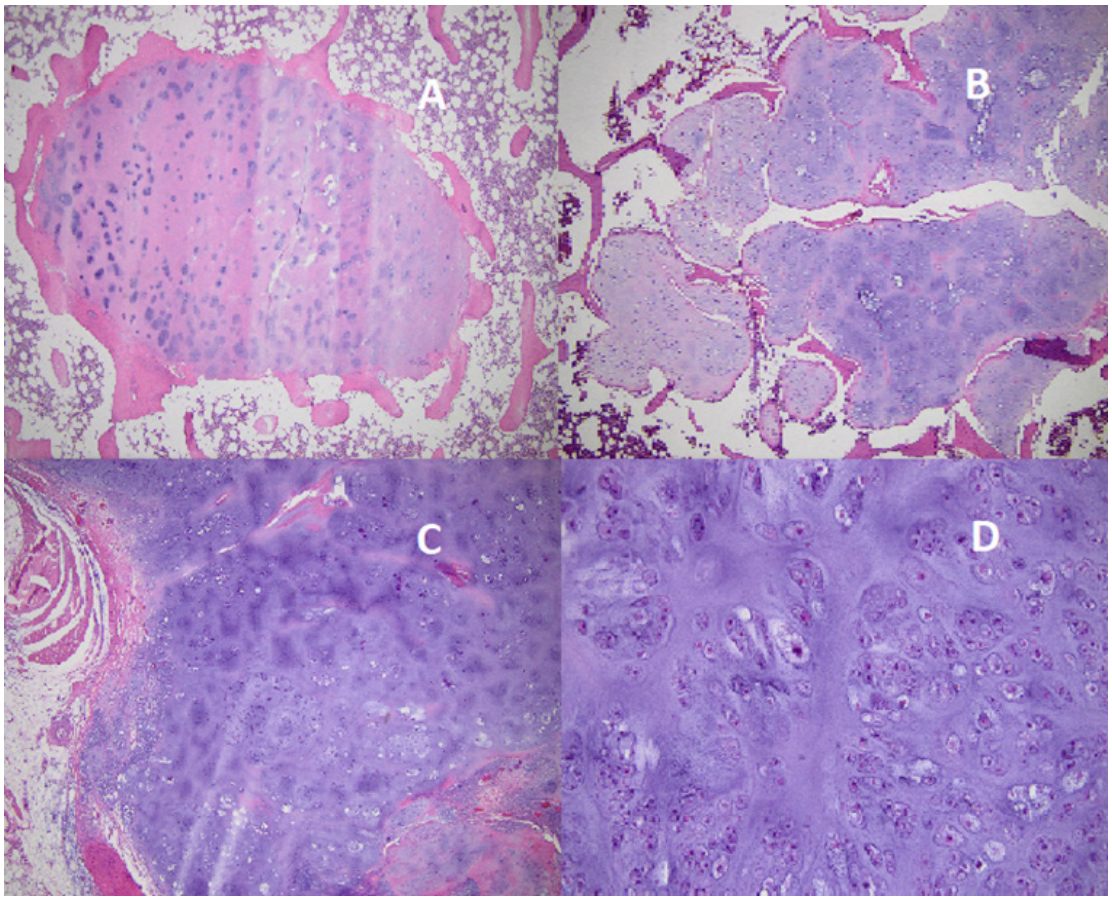

Fig. 2. Histological examination of the resection specimen, Hematoxylin eosin Ob4x

first week and triweekly in the following weeks (Figure 3). Treatment was well-tolerated, with Grade 2 radiodermatitis at the end of the treatment.

Discussion

Chondrosarcomas are the most common type of malignant tumor of the chest wall. They can be primary or secondary tumors that occur against the background of malignant degeneration of some enchondromas or osteochondromas. Of all chondrosarcomas, 25% are considered secondary chondrosarcomas [7]. Although primary chondrosarcomas of the chest wall are frequently diagnosed around the age of 40–60 years, secondary chondrosarcomas are usually diagnosed earlier, around the age of 25–45 years [8]. Most often, secondary chondrosarcomas of the thoracic wall are located anteriorly, at the level of the costochondral junctions. We noticed this in our case [9]. Chondrosarcomas can have three degrees of differentiation. Grade 1 is characterized by very rare atypical cells. Grade 2 is characterized by the presence of atypical cells but also normal cartilaginous cells, and Grade 3 is characterized by the predominant existence of atypical cells and rare normal cartilaginous cells [10]. Secondary chondrosarcomas have a better prognosis and have a much lower recurrence rate than primary chondrosarcomas [11].

Because enchondromas are often asymptomatic, diagnosis is often made with the help of imaging investigations. The radiological examination frequently reveals osteolytic lesions in the affected bones, relatively well-delimited by the surrounding bone tissue [12]. CT examination also reveals osteolytic lesions. At the same time, examination by nuclear magnetic resonance imaging (MRI) in these patients shows lobed lesions with an intermediate signal intensity in the T1 sequences and increased signal intensity in the T2 sequences. However, identifying enchondromas with sarcomatous degeneration based solely on imaging investigations is particularly difficult [13]. In this case, the suspicion of a malignant process was raised due to the osteolytic lesion at the level of rib 4, so the surgery consisted of the resection of three ribs. At the same time, given the current oncological guidelines, we did not consider that a preoperative biopsy should be performed, the osteolytic lesion being specific to malignant lesions.

Of all imaging investigations, MRI has the best specificity and sensitivity in the diagnosis of malignant degeneration of enchondromas. The main imaging aspects recorded in these cases are bone edema, periosteal reaction, and soft tissue invasion around the tumor [14]. In our case, no MRI could be performed in the emergency condition. However, the osteolytic lesion in the chest wall documented by CT examination showed the need for surgery.

The etiology of enchondromas is not fully known. However, some studies have shown that patients with enchondromatosis have mutations in the PTHLH receptor and the PTH1R subtype. This is a receptor for parathyroid hormone. In these cases, the sensitivity of this receptor to parathyroid hormone decreases. This mutation has also been found in patients with single chondromas or chondrosarcomas [15,16].

Histologically, several types of enchondromas exist. These are classified into Ollier disease, Maffucci syndrome, metachondromatosis, genochondromatosis, spondyloenchondrodysplasia, cheirospondyloenchondromatosis, and dysspondyloenchondromatosis. Identifying the histological type of enchondroma is particularly important, in terms of both the therapeutic decision to be made and identifying the genetic substrate of the disease [17].

Ollier disease, also known as dyschondroplasia, is the most common type of enchondromatosis and is characterized by multiple enchondromas, arranged asymmetrically

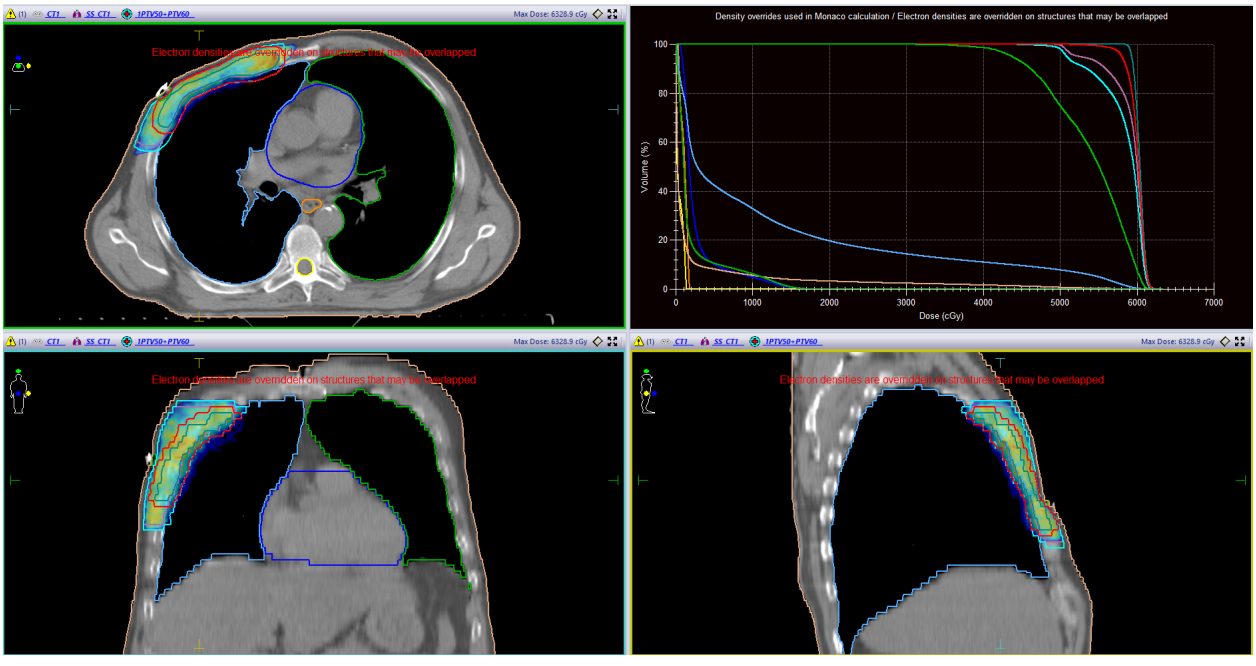

Fig. 3. Radiotherapy planning.

in the skeleton. It is most commonly diagnosed during childhood, with no difference in incidence between sexes [18]. It is not currently considered to have a genetic transmission, although some authors in the literature suspect this [19]. Studies have shown an increased risk of malignant transformation of enchondromas in these patients, as well as the risk of developing malignant tumors in tissues other than bone, especially nerve tissue [20].

Maffucci syndrome is characterized by the association of enchondromas with hemangiomas located in the soft tissues, as well as lymphangiomas [21]. It is frequently diagnosed in childhood, with hemangiomas often calcifying and cavernous or capillary [22]. It is considered a non-familial disease, with both enchondromas and hemangiomas at risk of malignancy in these cases. The risk of malignant degeneration of these lesions is believed to be higher than in Ollier disease [23].

Metachondromatosis is a genetically transmitted, autosomal dominant disease characterized by osteochondromas and osteochondroma-like lesions in the same patient. Most commonly, enchondromas are located in the long bones of the lower limbs, as well as the iliac crest, and osteochondromas are usually located in the small bones of the foot and hand [24]. No cases of malignant degeneration of these lesions have been reported in the literature, only cases of avascular necrosis of the femoral head [25].

Genochondromatosis is a genetic condition with autosomal dominant transmission, characterized by enchondromas arranged symmetrically in the proximal metaphysis of the humerus, as well as the distal metaphysis of the femur. It is usually diagnosed during childhood and does not cause deformity of the affected bones. No cases have been published attesting to the possibility of malignant transformation of these lesions [26].

Spondyloenchondrodysplasia is a genetic condition with autosomal recessive transmission, characterized by enchondromas in the long bones of the limbs and the pelvic bones, which are associated with vertebral dysplasia. It is usually diagnosed during childhood [27]. It is often associated with abnormalities of the nervous system, especially the spinal cord. No data have been cited in the literature attesting to the malignant degeneration of these lesions [28].

Cheirospondyloenchondromatosis is characterized by enchondromas in the metacarpal bones, as well as the phalanges of the lower and upper limbs. It is usually diagnosed during childhood and leads to deformity of the affected limb segments [29]. Dysspondyloenchondromatosis is characterized by enchondromas in the bones of the limbs, hands, and feet, which are associated with deformities of the spine. It is a condition without hereditary transmission, and no cases of malignant degeneration of these lesions have been cited in the literature [30].

In the case of unique, asymptomatic enchondromas that do not cause cosmetic defects, many authors recommend only clinical supervision, surgery being necessary only in case of enlargement of the lesions [31]. However, due to the risk of malignant degeneration of enchondromatosis lesions, treatment consisting of surgical resection of the lesion is indicated [32].

The only treatment that can cure patients with chondrosarcomas is surgery. Chondrosarcomas are usually resistant to chemotherapy and radiation therapy. Surgical resection should be performed within oncological safety limits, and the resection margins should be more than 2 cm from the tumor boundary. If more than three ribs are resected, or if the size of the thoracic parietal defect exceeds 10 cm, to avoid instability of the thoracic wall or herniation of the lung at the level of the thoracic parietal defect, chest wall reconstruction procedures are recommended [33]. Chest wall reconstruction can be performed with either autologous materials such as musculoskeletal flaps (large dorsal muscle, anterior dentate muscle) or heterologous materials (polypropylene mesh, mesh composites) [34].

Resistance to adjuvant therapy is mainly due to poor vascularization of the tumor, the low rate of proliferation of neoplastic cells, and the dense hyaline extracellular matrix [35,36]. During their evolution, chondrosarcomas most frequently metastasize to the lung [37]. According to the literature, the five-year survival rate of these patients is around 50–60%, and at 10 years, it is around 30–40% [38].

The particularity of the presented case is represented by the appearance of a secondary chondrosarcoma at the level of the thoracic wall on the background of the malignant degeneration of pre-existing enchondromatosis lesions, the patient being completely asymptomatic before diagnosis.

Conclusions

In patients with even asymptomatic chest wall enchondromas, periodic clinical evaluation of these lesions is required, given their risk of malignant degeneration.

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Authors' contribution

I.C.F. (Conceptualization, Methodology, Writing – original draft), I.I.C. (Project administration), A.G. (Formal Analysis), B.A.S. (Data curation), D.M. (Visualization), V.N. (Resources), A.A.M. (Validation), I.H. (Writing – review & editing)

Conflict of interest

The authors declare no competing interests

Informed consent

The patient signed an informed consent for the publication of this manuscript

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