

Hyperplasia of rib and vertebra, associated with infiltrating lipoma: a rare case of focal overgrowth

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Abstract

Purpose Syndromes with focal overgrowth are rare and diagnosis is difficult because manifestations are highly variable and symptoms overlap between syndromes. Diagnosis depends on clinical history, physical examination, and radiologic and histologic findings. This report describes a case of focal overgrowth of the left seventh rib and half of the adjacent thoracic vertebra, with overlying infiltrating lipoma.

Methods A 13-year-old boy presented with an asymptomatic chest wall mass caused by enlargement of the seventh rib and an overlying soft-tissue mass accompanied by enlargement of half of the seventh thoracic vertebra. MRI showed infiltration of lipomatous tissue in the muscles, but no interfascicular accumulation of adipose tissue in the thoracic spinal nerve.

Results A similar case was presented in 1985 but without MR imaging.

Conclusion We report on a second case of focal overgrowth of a rib and half of the adjacent vertebra, and overlying lipoma. In addition to the first case, we present MR images demonstrating infiltration of the adipose tissue.

Keywords Focal overgrowth · Nerve territory · Hyperplasia rib

Introduction

Focal overgrowth of bone associated with lipomas can be seen in various degrees and as part of multiple syndromes. Differentiation between these syndromes can be difficult due to the variability of symptoms.

Proteus syndrome and CLOVE syndrome show a mosaic distribution throughout the body [1, 2]. Other syndromes have more localized manifestations. These include macrodystrophia lipomatosa [3] and hemihyperplasia syndromes [4]. Most syndromes with overgrowth manifest in extremities. Rarely, cases of localized overgrowth in the chest wall have been described in the literature [5].

In this report, we describe the second case in the literature of focal hyperplasia of a rib and half of the adjacent vertebra associated with overlying lipoma.

Case report

A 13-year-old boy presented with a painless mass in the region of the left scapula, which had first been noted at the age of four. The mass had slowly increased in size during the subsequent years, but further remained asymptomatic. Past medical history included premature birth at gestational age of 33 2/7 weeks with a birth weight of 1,675 g as second child of twins. No history of teratogen exposure was known. He was in fetal distress immediately after birth, but had a furthermore uncomplicated neonatal period after that. At age five he was admitted to the hospital for failure to thrive, which was due to pleural effusion with unknown cause, and resolved spontaneously. He takes no medication.

Physical examination revealed a painless mass beneath the left scapula, a slight thoracic scoliosis and a prominent

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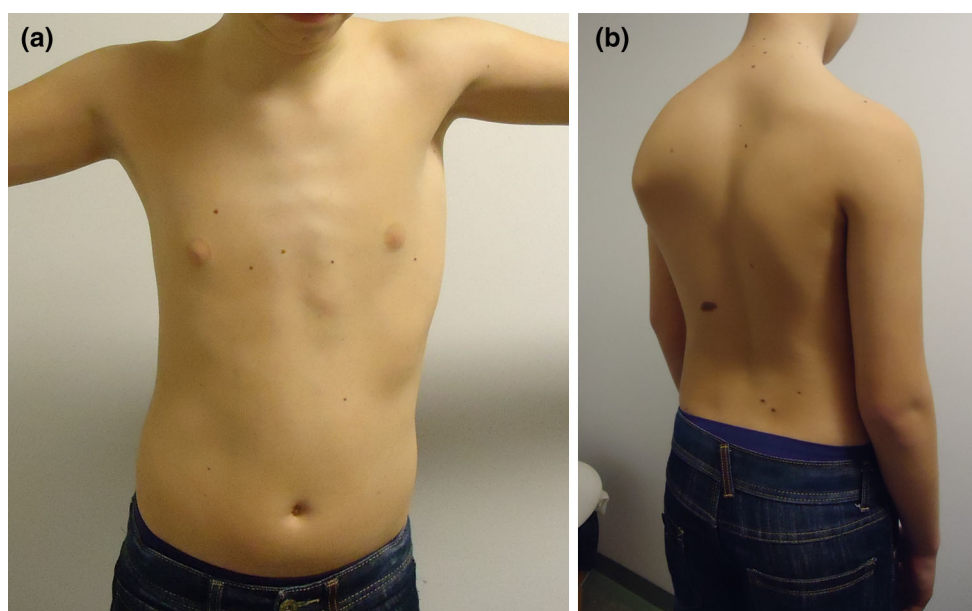


Fig. 1 Anterior (a) and postero-lateral (b) view of asymptomatic mass inferior to the left scapula and extending in the course of the seventh rib to the anterior side

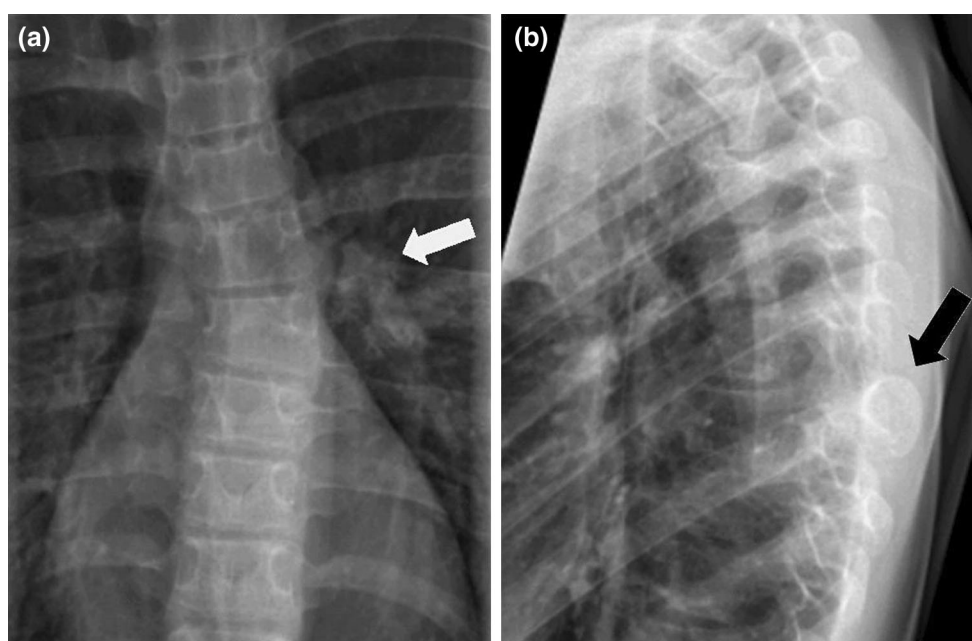


Fig. 2 Conventional radiographs of thoracic spine, showing a thoracic scoliosis, and enlargement of the seventh rib and half of the seventh thoracic vertebra. On the anteroposterior view (a) the

scoliosis can be appreciated and the enlargement of the costovertebral articulations (*light gray arrow*). On the lateral view (b): enlargement of the seventh rib (*black arrow*)

rib, predominantly visible on the anterior and lateral side (Fig. 1). There was no sensory disturbance over the affected area. The boy had one non-specific nevus on the left side of his back and had no vascular malformations over the body.

Conventional radiographs of the thoracic spine showed a left convex scoliosis with a 21° Cobb's angle, caused by

hyperplasia of the left side of the seventh thoracic vertebra, and hyperplasia of the seventh rib on the left side (Fig. 2). MRI demonstrated hyperplasia of the left side of the body and arch of the seventh thoracic vertebra and the seventh rib. The seventh rib had a distinct increase in volume compared to the adjacent ribs. Overlying are atrophic paraspinal muscles and a fatty tissue mass, infiltrating these

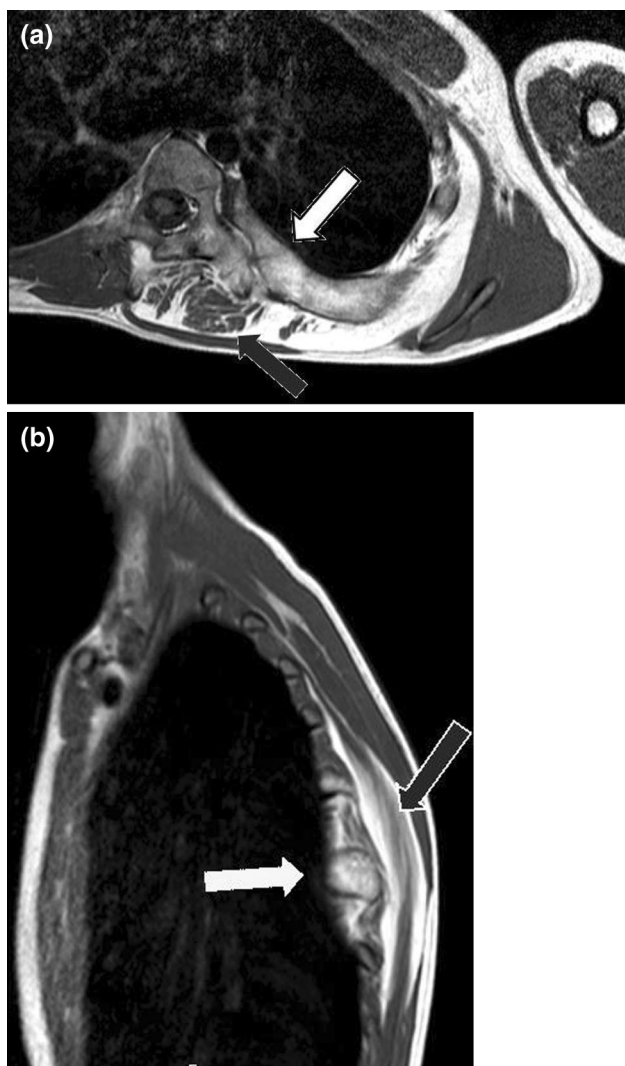


Fig. 3 Axial (a) and sagittal (b) T1-weighted MR images showing enlargement of seventh rib and adjacent half of the thoracic vertebra (light gray arrows). Overlying is a lipomatous mass infiltrating in the paraspinal muscles (dark gray arrows). Cutaneous and subcutaneous tissues and other thoracic muscles are normal

muscles (Fig. 3). There were no signs of organ hypertrophy or focal vascular malformations. Because the lesion was benign and asymptomatic no biopsy was performed.

Discussion

Syndromes with focal overgrowth are rare and diagnosis is difficult because manifestations are highly variable and symptoms overlap between syndromes. There are no genetic tests yet to diagnose a specific syndrome, so diagnosis is dependent on clinical history, physical examination, and radiologic findings [1]. In most descriptions of focal overgrowth, the overgrowth is localized in an extremity [1, 2].

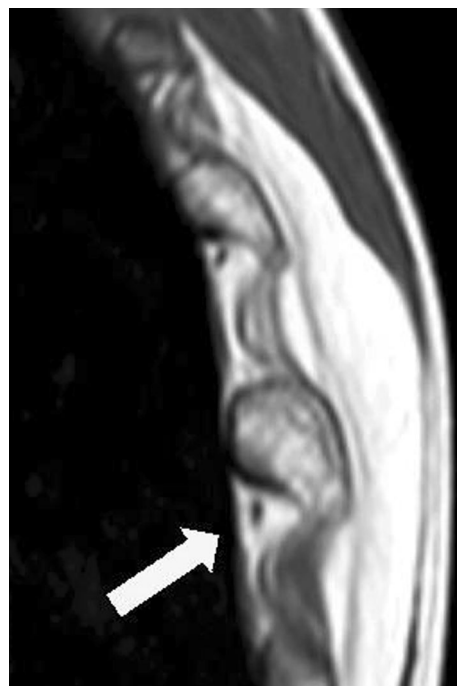


Fig. 4 Sagittal T1-weighted view of the dorsal part of the chest wall including the normal sixth and hyperplastic seventh left rib, showing the intercostal blood vessels (light gray arrow), and absence of thickening of the accompanying nerve by interfascicular infiltration of adipose tissue

In macrodactyly, Upton [6] uses a classification to differentiate between several kinds of focal overgrowth. He describes a true macrodactyly that is nerve-oriented and can be static (type Ia), or progressive (type Ib). Furthermore, he describes three types of pseudomacrodactyly with neurofibromatosis (type II), with hyperostosis (type III), and with hemihypertrophy (type IV).

The case presented here most resembles the type Ia macrodactyly. However, while it is not in the hand or foot, it may better be just termed focal overgrowth.

To our knowledge, overgrowth of a rib and dorsal vertebra has previously been described in the literature only once by Sauer et al. [5]. They showed the case of an 8-year-old boy who had an asymptomatic mass due to hyperplasia of the left sixth rib and left half of the adjacent vertebra. Overlying was mature adipose or lipomatous tissue without evidence of invasion into surrounding tissue. No MRI analysis was performed and the overlying tissue was removed. Histologic examination showed the excised tissue to be normal. Pathogenesis of this rib overgrowth is still unknown, but Sauer hypothesized that the sensory nerve, after being affected by a teratogen or other agent, induces altered soft tissue and osseous development through a trophic influence, with resultant lipoma formation and dysostosis [5]. In our case no history of teratogens during embryonic development was known.

In true macrodactyly debate exists on the etiology of the condition. In macrodactyly of the hand, the nerve is predominantly enlarged in most cases [7], and this enlarged nerve is considered to be the cause of the overgrowth [8]. In macrodactyly of the foot, however, the nerve is often not enlarged, although the overgrowth follows the course of a peripheral nerve [7].

In Sauer's case the adipose tissue seemed to be noninvasive, not infiltrating surrounding tissues. In our case no tissue was removed. Therefore, histological examination could not be performed, but MRI-scan showed infiltration of fatty tissue in the paraspinal muscles. The hyperplastic tissues are all in the area of the left 7th thoracic spinal nerve. This relation between overgrowth and nerve innervation is also described in a spectrum of diagnoses, called lipomatosis of nerve. However, one of the features of lipomatosis of nerve is intraneural deposition of adipose tissue [9], which can easily be detected on MRI images as fascicles in a nerve which are separated by adipose tissue leading to thickening of the spinal nerve. This feature is not present in our case (Fig. 4).

Although the etiology is still unknown, we can conclude that the presented case shows a second case of focal overgrowth of a rib with half of the adjacent vertebrae and overlying lipoma.

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Conflict of interest None.

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