

## CASE REPORTS

# Rhabdomyomatous Mesenchymal Hamartoma

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**Abstract.** Rhabdomyomatous mesenchymal hamartoma is an extremely rare congenital lesion, and very few cases have been reported even though its macroscopic and microscopic features make diagnosis easy. An 18-year-old woman consulted with a pedunculated mass in the medial region of her neck. The mass was surgically removed, and rhabdomyomatous mesenchymal hamartoma was diagnosed. The clinical, macroscopic, histologic, and immunochemical characteristics that allow diagnosis of this entity are discussed. Although association with congenital abnormalities is uncommon, this possibility should be assessed by the clinician.

**Key words:** skin abnormalities, cutaneous tumors, skin and connective tissue disease, skeletal muscle.

### HAMARTOMA MESENQUIMAL RABDOMIOMATOSO

**Resumen.** El hamartoma mesenquimal rabdomiomatoso es una lesión congénita extremadamente rara, con características macro y microscópicas que ayudan a su fácil diagnóstico; a pesar de lo anterior, se han descrito muy pocos casos. Presentamos el caso de una paciente de 18 años de edad, con una masa en la región medial de cuello, pediculada, la cual se extirpó quirúrgicamente obteniendo el diagnóstico de hamartoma mesenquimal rabdomiomatoso. Se analizan las características clínicas, macroscópicas, histológicas e inmunohistoquímicas, que permiten diagnosticar esta entidad. La asociación con anomalías congénitas, aunque poco frecuente, debe ser valorada por el clínico.

**Palabras clave:** anomalías cutáneas, neoplasias cutáneas, enfermedades de la piel y tejido conjuntivo, músculo esquelético.

## Introduction

Rhabdomyomatous mesenchymal hamartoma is a benign congenital lesion that was first described in 1986 by Hendrick,<sup>1</sup> although it was not until 1989 that the term itself was used for the first time by Mills.<sup>2</sup> In 1990, Elgart and Patterson<sup>3</sup> used the more general term congenital midline hamartoma, and it is also known as rhabdomyosarcomatous mesenchymal hamartoma,<sup>4</sup> striated muscle hamartoma,<sup>5</sup> and hamartoma of cutaneous adnexa.<sup>6,7</sup> It occurs as single or multiple cutaneous lesions that are often polypoid and typically appear on the midline,<sup>4-6</sup> and which have distinctive histologic characteristics. They are composed of mature striated

muscle fibers running vertically in relation to perilesional skin and display focal penetration of the dermis; they also contain mesenchymal elements such as adipose, connective, vascular, and nerve tissue, and skin adnexal structures.<sup>6,8</sup> We report a case of this lesion.

## Case Description

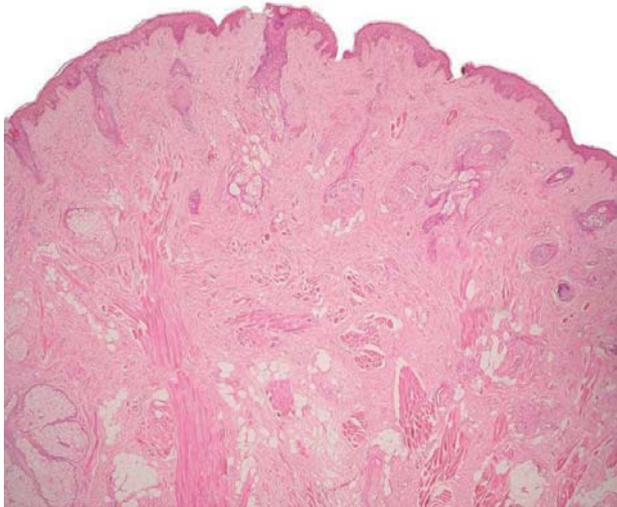
The patient was an 18-year-old woman who presented with a pedunculated mass on the medial area of the neck. She said that the mass had been there since childhood and that there had been no recent growth. The diagnostic impression was of a branchial cyst, which was surgically removed without complications.

The pathology department reported an elastic, polypoid, skin-covered mass measuring  $1.3 \times 0.8$  cm at its widest point. Incision revealed a tumor involving the dermis and subcutaneous tissue that was reddish brown and meaty in texture; there was no evidence of cysts. The tumor sample was fixed with 10% formaldehyde and stained with hematoxylin-eosin.

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**Figure 1.** Histologic section showing the polypoid nature of the lesion (hematoxylin-eosin).

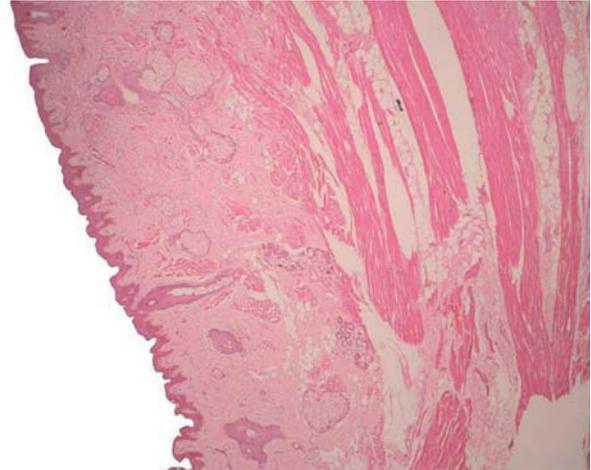


**Figure 2.** Apical segment of the lesion. Note the fine skin composed of epidermis and dermis; bundles of skeletal muscle arranged perpendicularly to the epidermis can be observed at the level of the reticular dermis (hematoxylin-eosin  $\times 4$ ).

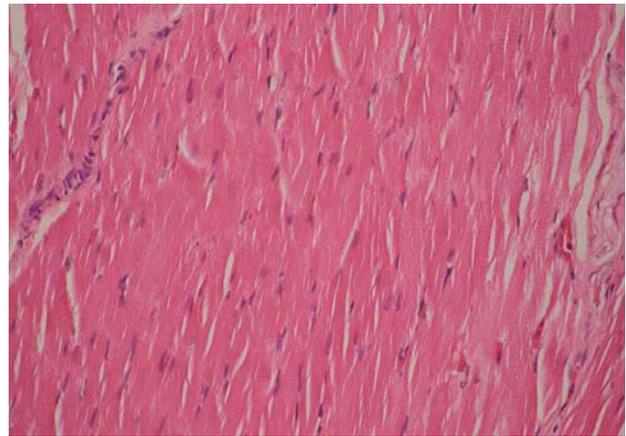
Histology revealed a benign mesenchymal polypoid tumor covered with epidermis and dermis and composed of skeletal muscle fibers, mature adipose tissue, and fibrous connective tissue arranged perpendicular to the surface of the adjacent skin and extending to the reticular dermis. The dermis contained hair follicles, sweat glands, and sebaceous glands (Figures 1, 2, 3, and 4). There was no evidence of malignancy.

## Discussion

Rhabdomyomatous mesenchymal hamartoma is one of the hamartomas reported in the region of the head and neck.<sup>6,9</sup> It usually presents as a polypoid, papular cutaneous lesion that is pedunculated or dome-shaped and ranges in size from a few millimeters to 1-2 cm.<sup>5</sup> The most common sites are the chin and nose, followed by the periorbital region, periauricular region, and anteromedial region of the neck.<sup>5,6,10,11</sup> Our literature search revealed 25 reported cases of this lesion, most of which were in newborns and children;



**Figure 3.** Lateral segments of the lesion. A benign mesenchymal polypoid tumor is visible covered with epidermis and dermis and composed of skeletal muscle fibers, mature adipose tissue, and fibrous connective tissue. The dermis contains hair follicles, sweat glands, and sebaceous glands. Hematoxylin-eosin,  $\times 4$ .



**Figure 4.** Detailed view showing the histology of the center of the lesion, where mature skeletal muscle is visible. Hematoxylin-eosin,  $\times 10$ .

only 3 were reported in adults (the eldest of whom was 54 years old).<sup>5,6</sup> They are twice as common in men as in women.<sup>12</sup> These lesions are composed of striated muscle that, on the basis of their anatomic distribution, is derived from the second branchial arch, since they are found in the region of the orbicularis oris, platysma, and orbicularis oculi muscles.<sup>6,7,8,12</sup> The etiology of this condition is unknown, although it may be due to an abnormality in the migration of mesodermal tissue during embryogenesis or to genetic defects<sup>12</sup>; it has been associated with other congenital defects,<sup>13</sup> such as gene defects (Disorganization mutation),<sup>12</sup> amniotic band syndrome, Delleman syndrome (also known as the oculocerebrocutaneous syndrome), and the Goldenhar syndrome, which includes colobomas, absence of the corpus

callosum, orbital cysts, porencephalic cysts, skin tags, and other abnormalities that do not form part of a known syndrome at the time of diagnosis.<sup>6,12-15</sup> The condition has also been considered an X-linked disorder, thus explaining its greater frequency in men.<sup>8,12</sup> Nevertheless, the cutaneous lesions reported in these syndromes often lack a suitable histopathologic description that would allow us to establish relationships between them; therefore, familial presentation of this lesion has not been reported.<sup>12</sup> Most are described as papules or polyps,<sup>5,10,13</sup> and some present as nodules<sup>6,16</sup> or sessile masses.<sup>14</sup> These neoplasms are almost always asymptomatic, painless, firm, and scarcely pigmented, do not change in size during their clinical presentation, and have the interesting property of contracting spontaneously or in response to autonomous stimuli.<sup>6,12</sup> The distinctive characteristic in microscopic analysis is the presence of skeletal muscle fibers inside the lesion arranged perpendicularly to the surface of the adjacent skin and surrounded by abundant collagen-rich connective tissue.<sup>1-16</sup> The lesion extends from the reticular dermis, which contains skin adnexa, particularly hair follicles, sweat glands, and sebaceous glands.<sup>15</sup> The presence of nervous tissue varies considerably, from a marked presence to none identifiable.<sup>4,5</sup> There have been reports of other elements such as elastic cartilage, foci of calcification, and the presence of ossification centers.<sup>5</sup> Immunohistochemistry shows that the skeletal muscle fibers are positive for actin, desmin, and myoglobin.<sup>12</sup> The differential diagnosis is made with fibroepithelial polyp, lipomatous nevus, and accessory tragus<sup>6,10,12</sup>; the location at the midline and the microscopic component of skeletal muscle enables a distinction to be made between these lesions. Other differential diagnoses to be borne in mind are primitive tumors such as fetal rhabdomyoma, fibrous hamartoma of infancy, and neuromuscular hamartoma (benign triton tumor).<sup>6</sup> The recommended treatment is local surgical resection, and no recurrences have been reported.<sup>8,12</sup>

In conclusion, we report a case of rhabdomyomatous mesenchymal hamartoma and analyze the clinical, macroscopic, histologic, and immunohistochemical characteristics that enable this entity to be diagnosed. Although uncommon, the link with congenital anomalies should be evaluated by the clinician.

#### Conflict of Interests

The authors declare no conflicts of interest.

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