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Case Presentation

Striated muscle hamartoma presenting as a chin cyst in a newborn

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Abstract

Striated muscle hamartoma is a rare, benign mesenchymal neoplasm that typically arises in the midline of a newborn patient. We report a clinically and histopathologically classic case of striated muscle hamartoma presenting as a chin cyst in a newborn female.

Keywords: Striated muscle hamartoma, rhabdomyomatous mesenchymal hamartoma

Introduction

Striated muscle hamartoma, or rhabdomyomatous mesenchymal hamartoma, is a benign skin lesion that commonly occurs in the midline of neonates. Striated muscle hamartoma was first described by Hendrick et al. in 1986 in two newborn infants and then renamed "rhabdomyomatous mesenchymal hamartoma" by Mills in 1989 [1,2]. Subsequent case reports of both striated muscle hamartoma and rhabdomyomatous mesenchymal hamartoma have noted associations with congenital anomalies as well as diagnosis in adult and pediatric patients. We report a case of striated muscle hamartoma presenting as a chin cyst, clinically thought to be a sebaceous cyst, in a newborn female.

Case synopsis

An otherwise healthy female infant, born at term, was noted to have an approximately 1 cm cyst, which appeared to involve the skin and subcutaneous tissue at the tip of her chin without erythema or drainage. An ultrasound demonstrated an oval-shaped, hypo-echogenic nodule in the subcutaneous fat superficial to the mentum, whose tip measured 2 x 1.3 mm. The patient remained asymptomatic but the growth did not regress. Because of its prominent location on the tip of the chin, it was quite disfiguring. It was therefore electively excised when the patient was one year of age.

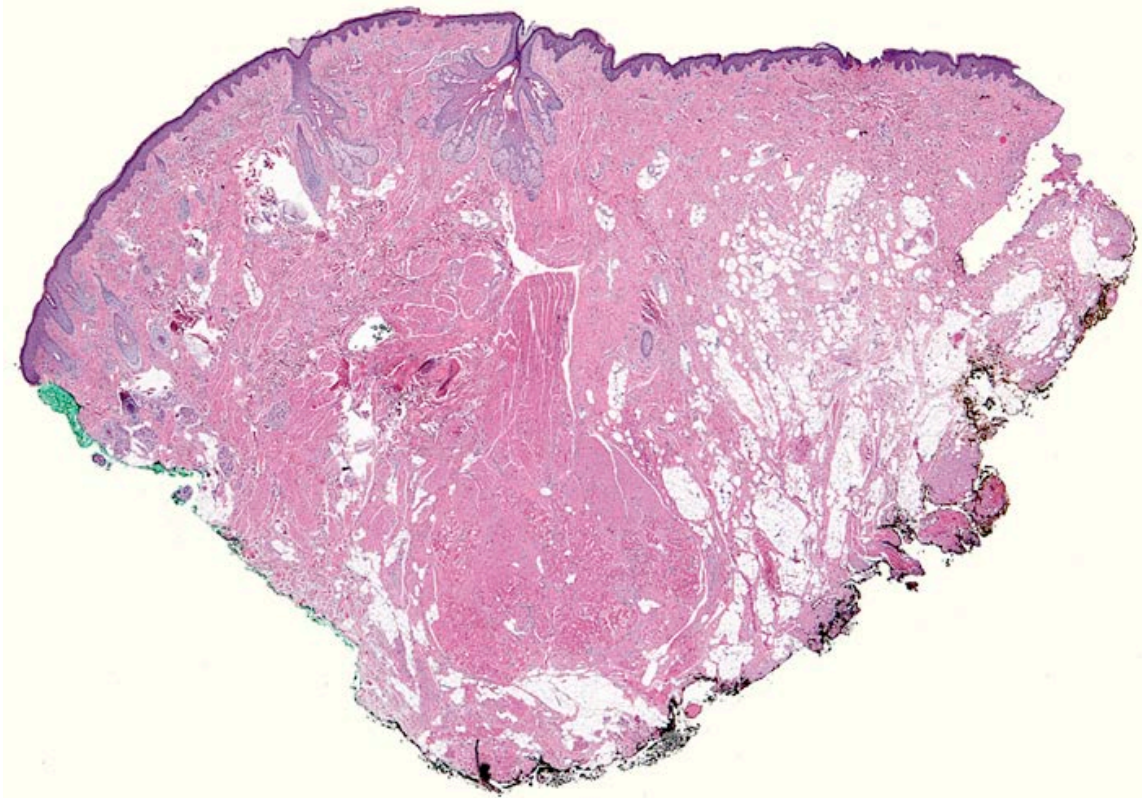
An excisional biopsy was performed. Intraoperatively, the nodule appeared to involve subcutaneous fat and muscle as well as skin at its tip. Gross evaluation demonstrated a tan, irregular piece of skin measuring 1.1 x 0.8 cm, excised to a depth of 0.5 cm. Cut surfaces were tan, smooth, and focally hemorrhagic.

Histopathologic sections of the chin biopsy demonstrate a hair follicle surrounded by a core of vertically-oriented skeletal muscle fibers. The muscle fibers appear normal and are surrounded by fibroadipose tissue. Unremarkable hair follicles, sebaceous glands,

and eccrine glands are also present. Trichrome and an immunohistochemical stain for desmin highlight the prominent striated muscle fibers. An EVG stain did not show an increase in elastin fibers.



Figure 1. Clinical image demonstrating the cystic-appearing lesion on the patient's chin.



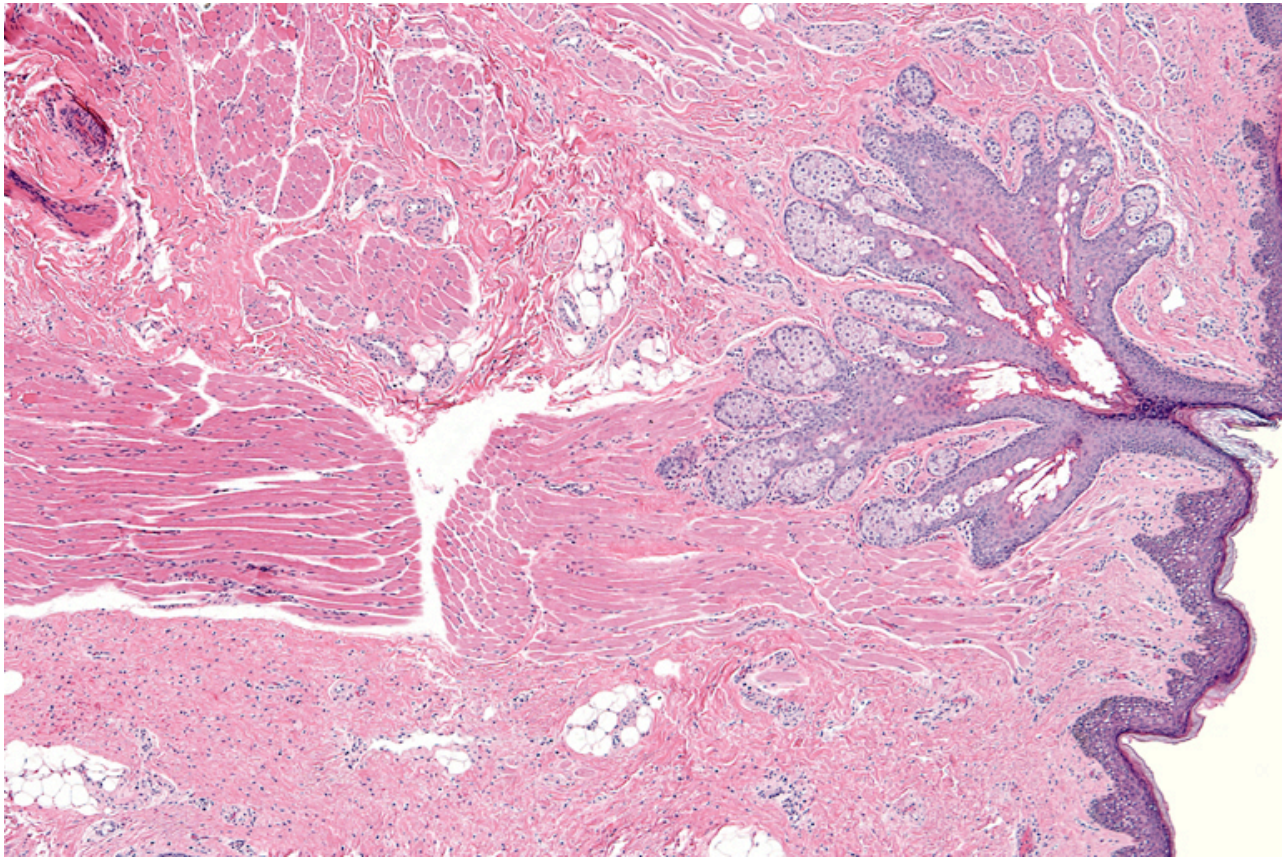
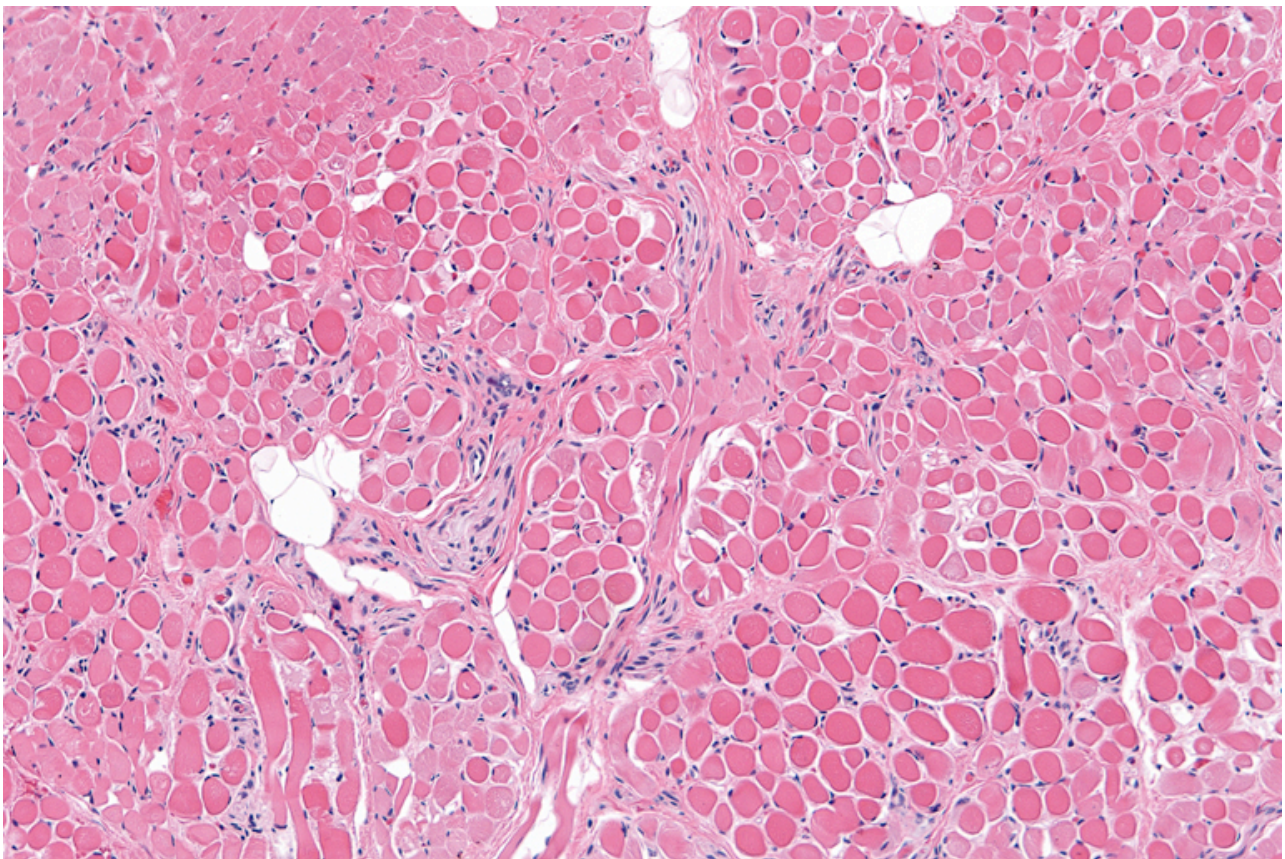


Figure 2. Histologic sections (2a: 1X, 2b: 4X) of the chin biopsy demonstrated hamartomatous bland skeletal muscle surrounded by fibroadipose tissue. Unremarkable hair follicles, sebaceous glands, and eccrine glands are also present.



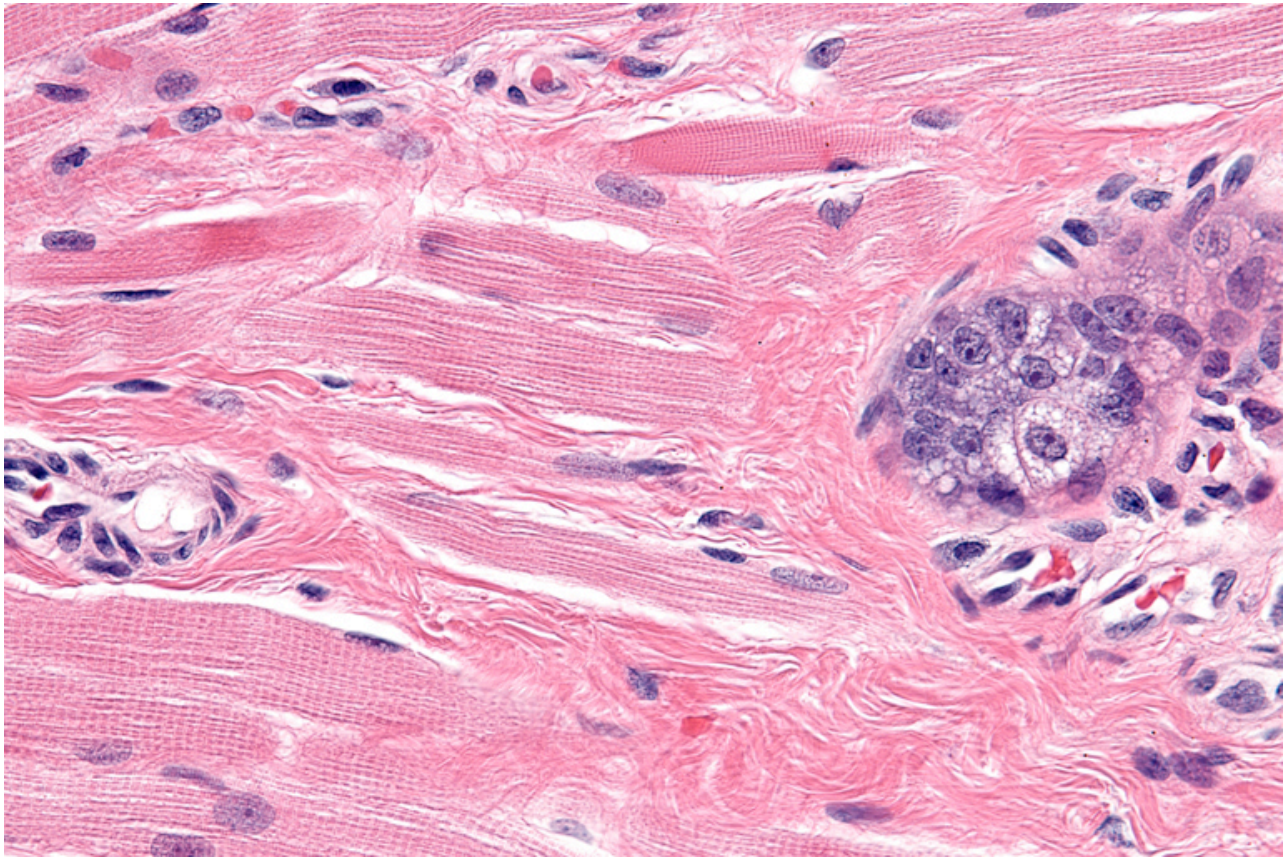


Figure 3. High magnification of striated muscle fibers in cross section (3a: 10X) and adjacent to adnexal structure (3b: 40X)

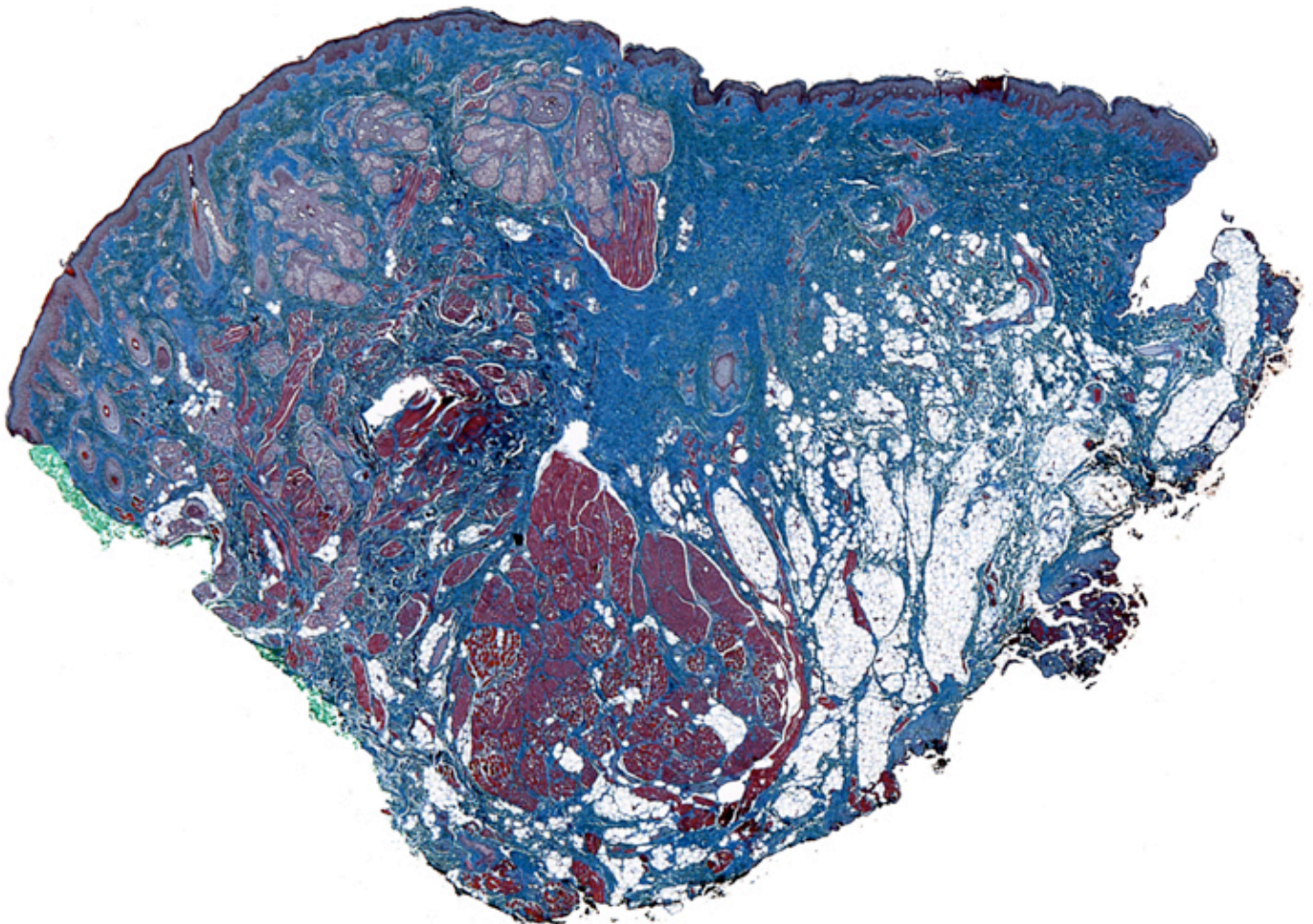


Figure 4. A trichrome stain highlights the prominent vertically-oriented skeletal muscle fibers in the dermis.

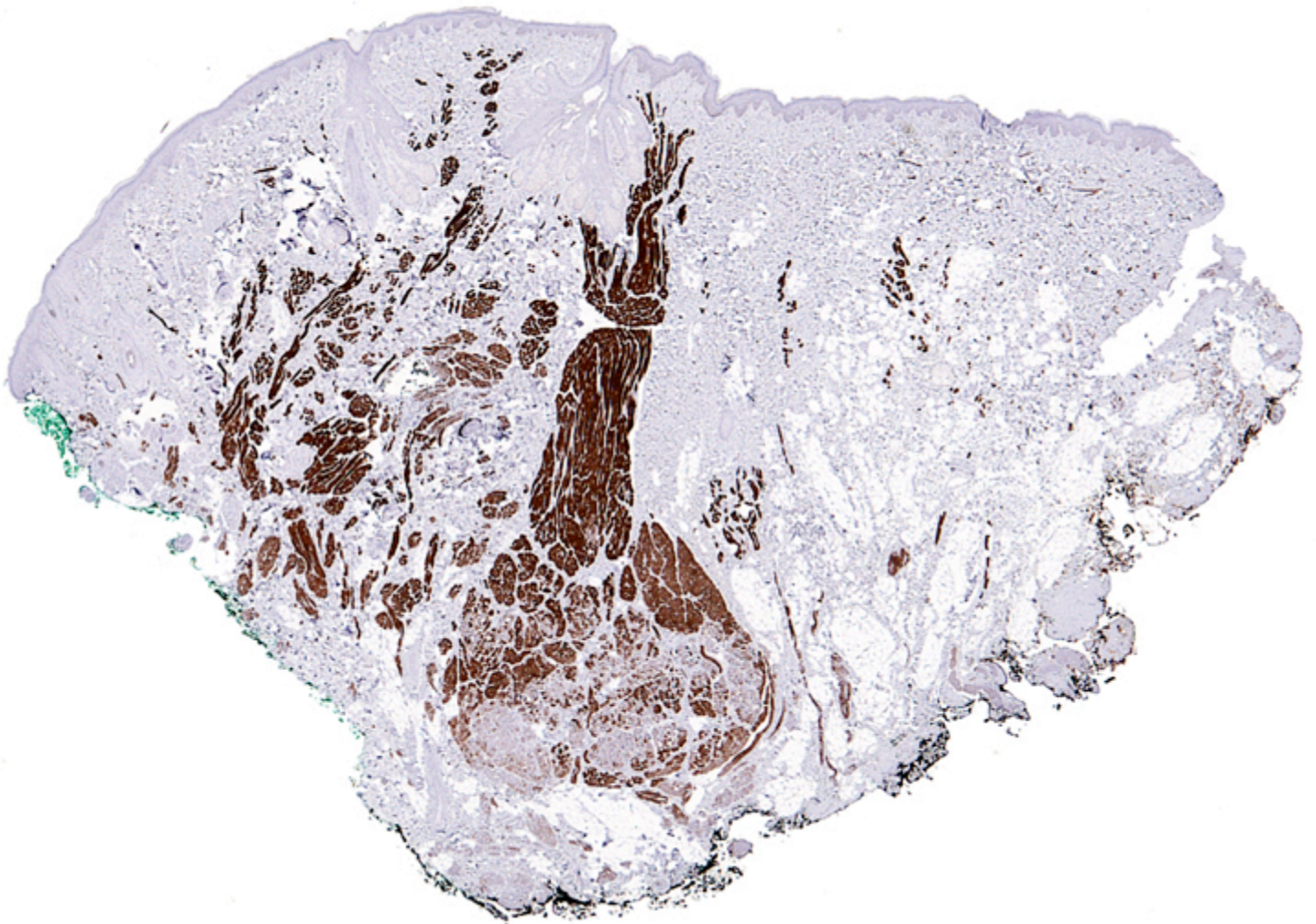


Figure 5. Immunohistochemistry for desmin highlights the prominent skeletal muscle fibers in the dermis.

Discussion

The clinical presentation and histopathological findings are consistent with striated muscle hamartoma. These growths occur in both males and females, are typically present at birth, and occur in the midline [1–10]. Whereas lesions often occur on the head and neck, midline lesions of the vagina and perianal area have also been reported [11,12]. Unusual cases describing presentation as multiple nodules [13–16], in non-midline anatomic positions such as the great toe [17], or in older children and adults have also been described [3,17–20].

This hamartoma typically presents as a nodule or skin tag, but has also been described as a solitary indurated plaque in a 42-year-old female and as depressed plaques resembling scleroderma “en coup de sabre” in a 17-year-old female [19,21]. A number of case reports have noted an association with additional congenital abnormalities and with Delleman syndrome [1,13,22,23]. Striated muscle hamartoma is a benign disorder without reported metastasis.

The diagnosis of striated muscle hamartoma is based on the presence of mature skeletal muscle fibers and other mesenchymal elements (fat, blood vessels, nerves). The dermis typically shows numerous cutaneous adnexae, including hair follicles, sebaceous glands, and sweat glands. Prominent bundles of striated muscle fibers are often seen perpendicular to the epidermis and running alongside sebaceous glands [1,2]. The epidermis is usually unremarkable. However, a case of lentiginous junctional melanocyte proliferation overlying rhabdomyomatous mesenchymal hamartoma has been reported [24]. Rhabdomyoma is an important consideration in the differential diagnosis.

This case highlights a clinically and histopathologically classic example of a rare mesenchymal neoplasm, striated muscle hamartoma, in the midline of a newborn. Histopathologic evaluation is necessary to make this diagnosis because the clinical presentation of the hamartoma can be quite variable. Increased awareness of this unusual entity is necessary to avoid misdiagnosis and overtreatment.

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