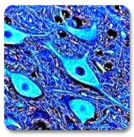

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Case report **Open Access**

Rhabdomyomatous mesenchymal hamartoma (RMH) of the anal region: an unusual location for such a rare lesion

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Abstract

Rhabdomyomatous mesenchymal hamartoma (RMH) is a rare tumour-like lesion, which occurs in the skin of newborns, principally of the face and neck. RMH has also been reported in unusual sites, including oral cavity, nasal vestibule and vagina. In this case report we describe a rare case of RMH arising as a nodular mass at the anal margin of a male newborn. This is an unusual site for such a rare lesion. The histological examination of the lesion showed the typical features of RMH. Desmin and S100 expression has been determined by immunohistochemical analyses to assess the distribution of the mature skeletal muscle cells and small-sized nerves, which represented the principal components of the nodular mass. Our case emphasizes the possibility that RMH may occur in the perianal region and awareness of this possibility is crucial for pathologists to avoid confusion with other skeletal muscle-containing lesions, especially rhabdomyosarcoma.

Keywords: Rhabdomyomatous mesenchymal hamartoma, nodular mass, perianal region, immunohistochemistry

Introduction

Rhabdomyomatous mesenchymal hamartoma (RMH) is a raretumour-like lesion, which occurs in the skin of newborns, principally of the face and neck [1-6]. The term "hamartoma", first coined by Mills in 1989 [1], has been histologically proven, being this lesion composed of a disordered mixture of mature adipose and skeletal muscle tissue, adnexal structures and sometimes blood vessels and nerves [4]. Although RMH is usually a solitary lesion, it can present as multiple lesions [7], rarely in association with uncommon congenital abnormalities [8,9]. Although RMH is a lesion, which typically occurs in the newborns, some cases have been reported as congenital lesions [10,11] or diagnosed in adult patients [12-14]. Apart from the skin of the head and neck region, RMH has also been reported in unusual sites, including oral cavity [15-17], nasal vestibule [18] and vagina [19]. Clinically, it can present as small dome-shaped papule or a polypoid pedunculated lesions [1-6,15-19]. Notably, rare cases of RHM may undergo spontaneous regression [20,21]. The etiology of RMH is still unclear, but it is likely that an aberrant embryological development or migration of mesodermally derived tissues due to micro-environmental modifications and/or genetic factors may play a pathogenetic role.

We report a rare case of RMH arising as a nodular mass at the anal margin of a male newborn. This is an unusual site for such a rare lesion, with only two cases previously reported in the peri-anal skin [8,22].

Case presentation

A 9-month-old male presented with a single dome-shaped nodule, measuring 2.5 cm in greatest diameter, arising at the anal margin. Clinical history was unremarkable. The lesion was surgically excised. After two years of clinical follow-up, no local recurrence was observed. Histological examination (Haematoxylin and Eosin staining) showed the typical features of RMH: a nodular mass, covered by normal-appearing epidermis, composed of a disordered admixture of dermal fascicles of mature skeletal muscle fibers, haphazardly intermingling with fibrous tissue, small islands of mature adipose tissue, adnexal glands, blood vessels of venular type, and small-sized nerves

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(**Figures 1A** and **2A**). No cytological atypia, necrosis or mitosis were observed. Although the skeletal muscle cells and small-sized nerves were easily recognizable at light microscopy, immuno-histochemical analyses were performed using the standard streptavidin–biotin labeling technique (LSAB kit-Dako, Glostrup, Denmark). As expected, anti-desmin and anti-S100 protein antibodies were helpful in highlighting, respectively, all the skeletal muscle cells and small-sized nerves haphazardly distributed throughout the dermis (**Figures 1B** and **2B**).

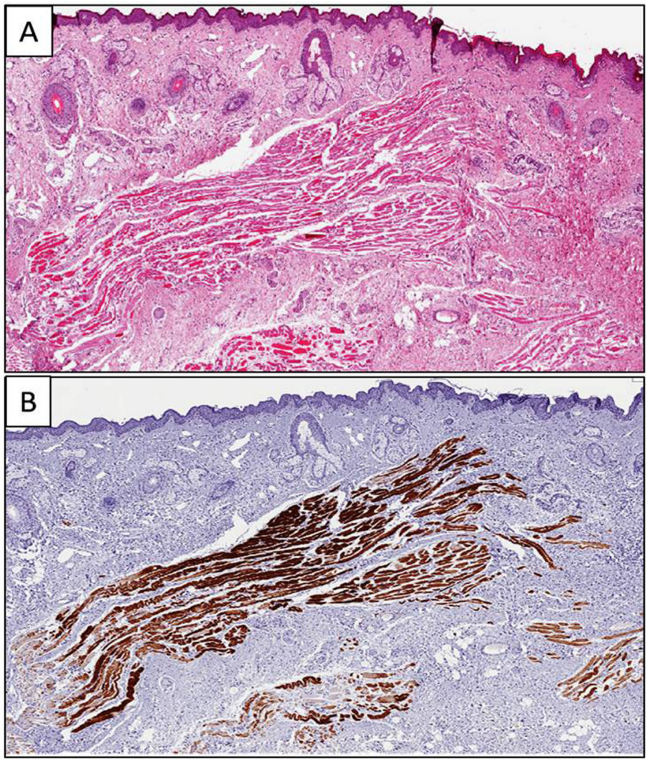

Figure 1. **Histological examination (Haematoxylin and Eosin staining)**.
(**A**) Low magnification showing a dermal lesion composed of fascicles of mature striated muscle cells scattered within the dermis and admixed with fibrous tissue, folliculo-sebaceous structures, rare adipocytes, blood vessels and small-sized nerves (haematoxylin and eosin staining; original magnification x60).
(**B**) Serial section stained with anti-desmin antibodies: the striated muscle cells exhibit a strong and diffuse cytoplasmic immunoreactivity (immunoperoxidase staining; original magnification x60).

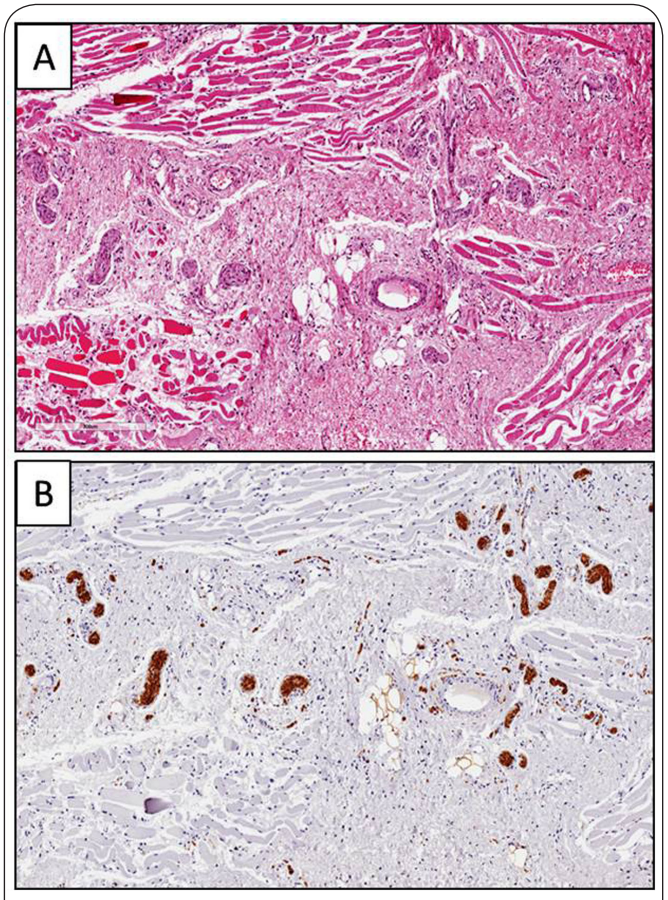

Figure 2. **Immuno-histochemical analyses using standard streptavidin-biotin labeling technique**.
(**A**) Higher magnification better showing the single components of the hamartomatous lesion: at the periphery are easily recognizable brightly eosinophilic striated muscle cells, while at the center of the lesion there is fibrous tissue in which are set small-sized nerves, blood vessels, and small islands of mature adipose tissue (haematoxylin and eosin staining; original magnification x100).
(**B**) Serial section stained with anti-S-100 protein antibodies: Schwann cells of the nerves exhibit a strong and diffuse cytoplasmic immunoreactivity (immunoperoxidase staining; original magnification x100).

Discussion

The present case shows that RMH may occur at the anal margin, and that this possibility should be considered preoperatively by clinicians, as well as by pathologists when dealing with a pediatric ano-rectal lesion. Although the two cases of perianal RMH reported in the literature were described as having a polypoid configuration [**8**,**22**], interestingly our case presented clinically as a dome-shaped nodule. Differential diagnosis of RMH mainly revolves around fetal rhabdomyoma and embryonal rhabdomyosarcoma. Fetal rhabdomyoma may rarely occur at the perianal region [**23**]. Among the different types of fetal rhabdomyomas, especially the intermediate (cellular) type may be confused with RMH. Infact the former contains numerous differentiated spindle-shaped striated muscle cells, but, unlike the latter, it lacks any additional mature tissue components (adipose tissue, adnexal glands, vascular structures, nerves) as integral part of the lesion. Embryonal rhabdomyosarcoma may present in the same sites, including perianal region, in which occurs RMH [**24**]. Unlike RMH, embryonal rhabdomyosarcoma shows a wide morphological spectrum of skeletal muscle differentiation,

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ranging from undifferentiated round to spindle-shaped, to well-differentiated mature cells exhibiting with cross-striations. In addition malignant neoplastic cells are usually embedded in a variably fibro-myxoidstroma with alternating hypocellular and hypercellular areas. All these features are lacking in RMH.

Conclusion

Our case emphasizes the possibility that RMH may occur in the perianal region and awareness of this possibility is crucial for pathologists to avoid confusion with other skeletal muscle-containing lesions, especially rhabdomyosarcoma.

Competing interests

The authors declare that they have no competing interests.

Authors' contributions

Authors' contributions	FL	GM	FA	FM	GM
Research concept and design	✓	✓	--	--	✓
Collection and/or assembly of data	✓	✓	✓	✓	✓
Data analysis and interpretation	✓	✓	✓	✓	✓
Writing the article	✓	--	--	--	✓
Critical revision of the article	✓	✓	✓	✓	✓
Final approval of article	✓	✓	✓	✓	✓
Statistical analysis	--	--	--	✓	✓

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