Rhabdomyomatous mesenchymal hamartoma presenting in a child as a perineal mass

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ABSTRACT

Rhabdomyomatous mesenchymal hamartoma (RMH) is a rare hamartomatous lesion in the dermis and subcutaneous tissue. It is mostly found in the face and neck region of children. We report a case of solitary RMH located in the perineum of an 8-month-old boy. Microscopic examination of specimen showed a disordered collection of mature adipose tissue, skeletal muscle, adnexal elements and nerve bundles, and immunohistochemistry confirmed a RMH. This case emphasizes the possibility of RMH in the perineum of the children. Even if RMH is a rare condition in the perineum it should be considered as a differential diagnosis of a perineal mass in children.

1. Introduction

Rhabdomyomatous mesenchymal hamartoma (RMH) is a rare, congenital, benign lesion of the dermis and soft tissue [1]. RMH usually occurs in areas with superficial striated muscle such as the chin and nose [2]. RMH was first described as a striated muscle hamartoma by Hendrick et al., in 1986 [3], officially named by Mills in 1989 [4], and the histopathological findings are unique [5]. The hallmarks of skin hamartomas are their congenital nature, their development within dermis, and the proliferation of mature striated muscles bundles associated with other mesenchymal structures. Differential diagnosis of RMH includes fibrous hamartoma of infancy (FHI), nevus lipomatosis superficialis, benign Triton tumor, fetal rhabdomyoma, and cutaneous embryonal rhabdomyosarcoma. Previously, only three cases of perianal RMH have been reported. In this case report we describe a case of solitary RMH located in the perineum of an infant.

2. Case report

An 8-month-old healthy boy was referred to our pediatric surgical center with an ill-defined perineal mass that had been present from birth. The 3.5 × 2 × 2 cm soft mass was located at the left side of the margin of the anal sphincter and it was asymptomatic (Figs. 1 and 2). Ultrasonographic scan revealed that the mass lied beside the anal sphincter. The decision was made to excise the lesion primarily to obtain diagnosis but also to prevent any future obstruction of passage of stool and also for cosmetic reasons. The surgery was performed when the child was eight months old, under general anesthesia with the child laying in prone position. There was no capsule of the tumor and it could be resected completely without any injury to the anus or anal sphincter. The tumor was sent for histopathological examination.

There was good symmetry of the buttock after suturing the skin. The child had normal defecation after the operation.

The histopathologic examination showed dome-shaped papule covered by normal stratified squamous epithelium. In the dermis and subcutis, tissue was seen as disordered collections of well-differentiated skeletal muscle fibers arranged aligned perpendicular to surface epithelium and interspersed with mixture of thick collagen bundles, small islands of mature adipose tissue, adnexal elements, blood vessels and nerve bundles (Fig. 3a and b).

No cytological atypia, necrosis or mitosis were observed. On immunohistochemical examination, mature skeletal muscle was positive...
Masson-Trichrome staining revealed mixed dark-red skeletal muscle and blue collagen bundles (Fig. 5).

Based on these clinical, histological and immunohistochemical findings, a diagnosis of RMH was made.

3. Discussion

Most of the patients with RMH in the medical literature were newborns or children under 3 years of age [6]. There have been no reports of malignant transformation occurring in RMH and no recurrences [7]. Spontaneous regression occurred in two patients [6,8]. Sex predilection is unclear, as some sources report a slight male predominance and others report no clear predominance [6,7]. The diagnosis is made during histopathology and the treatment is surgery.

To our knowledge, only three cases of perianal RMH have been reported. The first was in a 7-month-old female infant, noted when a large congenital perineal hemangioma regressed, revealing a polypoid mass at the anal margin [9]. The second was in a 3-month-old female infant with a congenital polypoid lesion in the perianal area [10]. The third case was a 3-month-old boy with a large perianal skin tag that had been present from birth. In all of the three cases the lesions were excised with no further recurrence [11].

RMH has been described as part of various syndromes such as Delleman syndrome (oculocerebrocutaneous syndrome), characterized by colobomas, orbital cysts, cerebral cysts, skin tags and an absent corpus callosum [12]. Possible associations with cleft lip/palate and amniotic bands syndrome have also been reported [3].

The etiology of RMH is still unclear. Possible hypotheses include aberrant embryonic migration of mesodermal tissue and the presence of a disorganized gene, found in mouse models with formation of limb defects, eye defects and hamartomas [13,14]. The presence of a RMH in patients with amniotic band syndrome has been theorized to be a result of traction caused by bands prompting the formation of RMH lesions [3]. A possible explanation for the unusual location and development of a RMH in our patient can be the proximity to the anal canal with the striated muscles of the pelvic floor and external anal sphincter.

FHI of infancy is a differential diagnosis of RMH and it most often occur at the posterior axillary fold. Histologically, it is a triphasic lesion, composed of an immature mesenchyme arranged in nests, concentric whorls or bands, an admixture of bundles of dense fibrous connective tissue, and mature adipose tissue. FHI does not contain skeletal muscle or nerves [15]. Nevus lipomatosis superficialis is another differential diagnosis that consists of the mature ectopic adipocytes within papillary or reticular dermis, thickened collagen bundles, scattered lymphocytes, and increased capillaries. Striated muscle fibers have not been reported [16]. Another differential diagnosis to consider is Benign Triton tumor or neuromuscular hamartoma that is composed of multiple nodules separated by narrow bands of connective tissue. Nodules are composed of fascicles of striated muscle fibers associated with peripheral nerve fibers (myelinated or not) within the same perimysial fibrous sheets [17]. Cutaneous embryonal rhabdomyosarcoma consists of a wide morphological spectrum of skeletal muscle differentiation, ranging from undifferentiated round to spindle-shaped, to well-differentiated mature cells with cross-striations [18].

4. Conclusion

Perineal RMH is a rare congenital, benign condition that should be considered in the differential diagnosis of anal lesions during childhood.
Fig. 2. Perianal rhabdomyomatous mesenchymal hamartoma preoperatively.
Patient consent

Consent to publish the case report was obtained from the parents.

Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Conflict of interest

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Appendix A. Supplementary data

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References


Fig. 3. a. Histopathologic examination (H&E, x 40). Overlying surface epithelium is unremarkable. Skeletal muscle fibers, seen within the dermis, tend to be aligned perpendicular to the surface epithelium. b. Histopathologic examination (H&E, x 100). The lesion consists of a disordered collection of mature adipose tissue, skeletal muscle, and adnexal elements.

Fig. 4. Immunohistochemical staining (desmin, x 40) showed the prominent skeletal muscle component.

Fig. 5. Histochemical staining (Masson-Trichrome, x 40) confirmed the ectopic presence of dark-red skeletal muscle mixed with blue collagen bundles.