Perianal rhabdomyomatous mesenchymal hamartoma

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ABSTRACT

A rhabdomyomatous mesenchymal hamartoma (RMH) is a rare benign soft tissue tumour found in the face and neck region of children. A three-month-old male infant was referred to our unit with a polypoid anal lesion that had been present from birth. Histopathological examination of the excised sample showed haphazard arrangement of mature striated skeletal muscle in among nerves and blood vessels, and immunohistochemistry confirmed the lesion to be a RMH. There was no evidence of recurrence during the follow-up period. This case emphasises two points: the unique histological characteristics associated with RMH and how this allows distinction from other differential diagnoses, and the consideration of RMH as a diagnosis in perianal lesions.

KEYWORDS

Rhabdomyomatous – Mesenchymal – Hamartoma – Paediatric – Perianal

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In 1989 Mills described a case of a solitary lesion on the chin of a neonate, which uniquely demonstrated the presence of striated muscle and mesodermal elements arranged in an abnormal pattern.¹ This rare histological finding was termed a rhabdomyomatous mesenchymal hamartoma (RMH). RMH is mainly described in children, and found most commonly as solitary lesions in the head and neck area. Cases of multiple lesions as well as those seen on the back, digits and mouth have also been reported. Perianal RMH has only been described in two cases. We report the case of an infant with a congenital incidence of RMH at the anal margin.

Case history

A three-month-old boy was referred to the surgical clinic with a large perianal skin tag that had been present from birth. The 1.7cm × 1.2cm polypoid mass lay distal to the margin of the anal sphincter at a 4 o'clock position and caused no symptoms. The decision was made to excise the lesion for cosmetic reasons and to aid diagnosis as well as prevent any future obstruction to passage of stool. This was done when the child was four months of age.

Histological examination showed a polypoid piece of tissue covered by stratified squamous epithelium. The core was composed of randomly arranged skeletal muscle, nerves and blood vessels. A subepithelial zone of loose connective tissue, fibrous connective tissue and scattered spindle cells (most likely myofibroblasts) was also seen (Fig 1). Antidesmin antibody highlighted the prominent skeletal muscle component and smooth muscle actin staining

showed vascular smooth muscle (Fig 2). There was very little proliferative activity identified by Ki-67 in the core of the polyp. The histological features were in keeping with a RMH. Reviewed again at four months following surgery, the child showed a good recovery with no recurrence.

Discussion

A hamartoma is a benign tumour-like malformation composed of normal cells but arranged in a haphazard pattern. Characterised by the disorganised arrangement of mature skeletal muscle, a RMH is a rare congenital malformation of the deep dermis and subcutaneous fat.² This condition is known under various names including striated muscle hamartoma, congenital midline hamartoma, and hamartoma of cutaneous adnexa and mesenchyme.³ These names are misleading as RMH contains more than simply striated muscle fibres and it has on occasion been found far from the midline.

The presentation of RMH is diverse as lesions can be polypoid or dome shaped, singular or multiple.^{4,5} The size of the lesions has been noted to range from 0.3cm to 1.4cm.⁶ The lesions are generally benign in otherwise healthy patients and are commonly only excised if causing mass effects or for cosmetic reasons.

The majority of RMH cases present in childhood although cases of adult presentation have been published. Time of diagnosis does not seem to affect histology findings. Indeed, some lesions excised in adulthood are reported as having been present throughout life.⁵

Figure 1 Histological examination of the polyp showing randomly arranged squamous epithelium, small spindle cells (myofibroblasts) in the subepithelial layer, and randomly arranged skeletal muscle, nerves and vessels.

Figure 2 Immunohistochemical staining for desmin showing the prominent skeletal muscle component.

The lesion arises from the dermis with a normal overlying epidermis. Characteristically, the histology shows a mixture of adnexal elements, with haphazard bundles of striated muscle fibres. Mesenchymal elements such as adipose tissue, blood vessels and elastic fibres are present in varying amounts.

The common differential diagnoses for RMH include nevus lipomatous superficialis (NLS), benign triton tumour, fetal rhabdomyoma, infantile myofibromatosis and smooth muscle hamartoma. Although both present in childhood, NLS and infantile myofibromatosis are histologically different from RMH. NLS is made up primarily of collagen bundles

and fat cells, and unlike RMH, no muscle fibres are seen.⁷ Infantile myofibromatosis presents as dermal and subcutaneous nodules in which myofibroblast derived spindle cells predominate. Triton tumours contain multiple cell types like RMHs but are associated mainly with nerve cells in the deep soft tissue.⁸ Foetal rhabdomyomas contain an immature striated muscle component whereas RMHs are made of mature striated muscle fibres.⁹ As the moniker suggests, smooth muscle hamartomas have a characteristically haphazard arrangement of smooth rather than striated muscle cells. In addition, they predominate on the chest wall or limbs. Given the variable presentation of RMH, histological investigation is the best tool for diagnosis.

RMH has been described as part of various syndromes such as Delleman syndrome (oculocerebrocutaneous syndrome), characterised by colobomas, orbital cysts, cerebral cysts, skin tags and an absent corpus callosum. In a case series by Sánchez and Raimier, 11 of the 12 patients with Delleman syndrome had singular lesions, which is histologically typical of RMH.¹⁰ Possible associations with cleft lip/palate and amnion band syndrome have also been reported.^{3,11}

The aetiology of RMH is still unclear. Possible hypotheses include aberrant embryonic migration of mesodermal tissue and the presence of a disorganised gene, found in mouse models with formation of limb defects, eye defects and hamartomas.^{4,12} The presence of a RMH in patients with amniotic band syndrome has been theorised to be a result of traction caused by bands prompting the formation of RMH lesions.¹¹

RMHs usually occur in areas with superficial striated muscle such as the chin and nose.¹³ Up until 2016, 46 cases had been reported in the English literature. An overwhelming number of cases (34 out of 46) involved lesions in the head and neck but appendages of the sacrum, digits and vagina have also been described.⁶ To our knowledge, only two cases of perianal RMH have been reported. The first was in a seven-month-old female baby, noted when a large congenital perineal haemangioma regressed, revealing a polypoid mass at the anal margin.¹⁴ The second was in a female infant aged three months with a congenital polypoid lesion in the perianal area.¹⁵ Both were excised with no further recurrence.

Although a rare presentation of a rare condition, Fröber *et al* describe the proximity of the anal canal to the striated muscles of the pelvic floor and external anal sphincter, possibly explaining the unusual location for RMH development.¹⁶ Diagnoses to consider in paediatric presentation of anal/buttock lesions include anal fissure, condylomata lata, NLS, polyp, granulomatous lesion of inflammatory bowel disease, infantile perianal pyramidal protrusion,¹⁷ acrochordon and rectal prolapse. As with all anal lesions, careful consideration of extension into the anal margin and muscle involvement is vital. The RMH anal lesions reported were all polypoid, with minimal extension, and were easily surgically excised with minimal disruption of the anal muscle complex. Novel methods for removal including laser therapy have been described but the instruments and expertise are not widely available.¹⁸

Conclusions

The large variety of possible diagnoses for anal lesions of childhood highlights the need for greater awareness of RMH as a differential diagnosis.

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