

Rhabdomyomatous Mesenchymal Hamartoma

Clinical Overview and Report of a Case with Spontaneous Regression

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ABSTRACT

A case of cutaneous rhabdomyomatous mesenchymal hamartoma in a 6-year old Afro-Caribbean girl is reported with review of the literature. The lesions were fine, located on the central face and became inapparent after six months. Spontaneous regression of these lesions has not been previously reported. Although rare, continued reporting will facilitate the elucidation of the clinical features and natural history of these lesions and the relationship to disordered embryogenesis.

Hamartoma Mesenquimal Rhabdomiomaso

Panorama Clínico y Reporte de un Caso con Regresión Espontánea

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RESUMEN

Un caso de hamartoma mesenquimal rhabdomiomaso cutáneo en una niña afrocaribeña de seis años de edad, se reporta junto con una revisión de la literatura. Las lesiones eran tenues, localizadas en la parte central de la cara, y se hicieron aparentes luego de seis meses. La regresión espontánea de estas lesiones no se ha reportado con anterioridad. Aunque sean raras, reportarlas de manera continuada facilitará la dilucidación de los rasgos clínicos y la historia natural de estas lesiones, así como su relación con una embriogénesis desordenada.

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INTRODUCTION

Rhabdomyomatous mesenchymal hamartoma (RMH) of the skin is a rare congenital malformation (1), first described as Striated Muscle Hamartoma by Hendrick *et al* in 1986 (2). Haphazardly arranged mature skeletal muscle fibres are central to the lesions which mainly affect the head and neck region, reportedly in males predominantly (3–5). Other mesenchymal elements may be admixed, resulting in a variety of other names including congenital midline hamartoma and hamartoma of cutaneous adnexa and mesenchyme. Fewer than 25 cases had been reported in the English literature up to 2002 (6) and only occasional case reports have followed subsequently. They may be solitary, independent lesions or multiple and associated with other congenital abnormalities (2, 4, 6–8). A case of RMH with spontaneous clinical regression in a 6-year old female child diagnosed at the University Hospital of the West Indies, Kingston, Jamaica

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is reported and the salient clinical features discussed. To our knowledge, this is the first such report from the Caribbean.

CASE REPORT

A 6-year old Afro-Caribbean girl presented to the Dermatology clinic at the Bustamante Hospital for Children (BHC) with fine papules on the central area of her face. Some of the lesions exhibited Koebnerization and were initially thought to be plain warts. She was treated with 3% salicylic acid in aqueous cream but there was no resolution. No other lesions were evident elsewhere. On review six-weeks later, the papules persisted and she was given imiquimod cream to be applied three times weekly but the lesions still had not resolved on review one month later. Electrocautery was attempted and repeated three months later without effect. Two months later, eight months after initial presentation, the lesions appeared less warty and were becoming more nodular. The differential diagnoses entertained were sarcoidosis and facial Afro-Caribbean eruption (FACE). A biopsy was taken from the left temporal area and she was commenced on 1% hydrocortisone cream with 2% precipitated sulphur. On review a week later, the lesions were flatter although itchy at times and she was treated with Desonide lotion at night and ketoconazole cream with Desonide lotion twice daily. At six

months follow-up, all lesions had cleared with no visible abnormalities and she was therefore discharged from the clinic.

Histopathological evaluation of the biopsy specimen revealed a somewhat polypoid 0.5 x 0.2 cm ellipse of skin. The overlying epidermis was unremarkable but the dermis contained bundles and individual fibres of skeletal muscle, haphazardly dispersed within lobules of mature adipose tissue separated by intersecting bands of fibrous tissue (Fig. 1a). The muscle fibres were of varying thickness and often

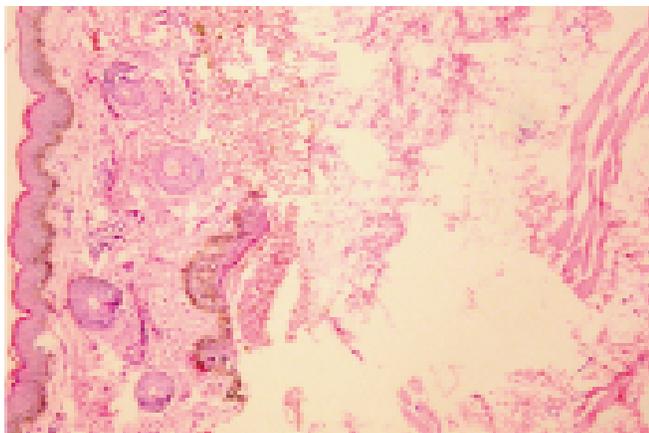


Fig. 1a: Low power skeletal muscle bundles in fibro-adipose tissue of the deep dermis (H&E).

were oriented perpendicular to the surface epidermis (Fig. 1b). Occasional nerve bundles and lobules of adnexal glands were also admixed (Fig. 1c).

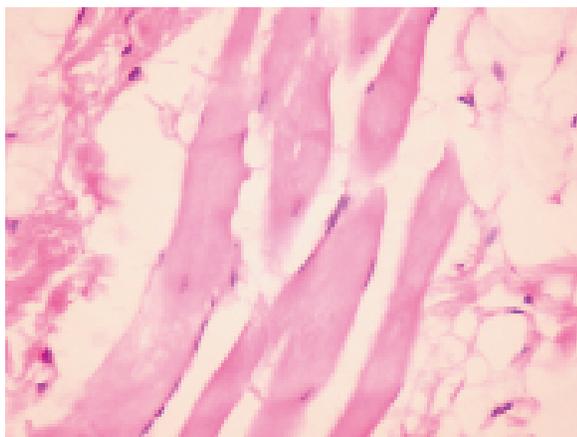


Fig.1b: High power of skeletal muscle fibres (H&E).

DISCUSSION

Many types of hamartomatous lesions may affect the skin (1–3, 6, 7, 9). When a myomatous component is present, this is usually smooth muscle. Striated muscle is distinctly uncommon and lesions with striated muscle component include fetal rhabdomyoma characterized by immature fetal

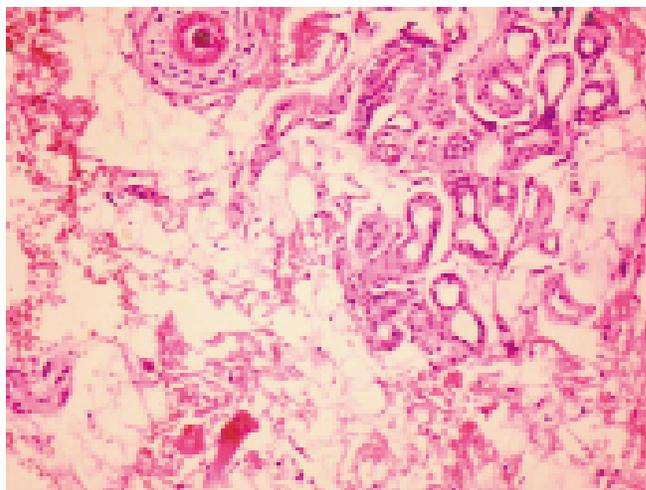


Fig. 1c: Low power of haphazard admixture of adnexal structures and scattered skeletal muscle fibres (H&E).

striated muscle admixed with undifferentiated mesenchyme and benign triton tumour which occurs associated with peripheral nerve in deep soft tissue and in which myelinated and unmyelinated nerves are admixed with the striated muscle fibres (1, 3, 6). The presence of mature skeletal muscle is central to rhabdomyomatous mesenchymal hamartoma. These muscle fibres are haphazardly dispersed within a variable lipomatous and fibrous stroma with a tendency to parallel orientation to the epidermis. This was the characteristic feature in the biopsy from this patient (Fig. 1a and 1b) although the gross clinical appearance was not typical. A variety of adnexal tissues such as eccrine glands may be admixed (Fig. 1c) and vellus hairs may also be present (2, 6, 7, 10). The significance of the adnexal component is debatable and may just reflect coincidental location in the dermis (10). Nevertheless, Rosenberg *et al* argue that coincidence is difficult to reconcile in large finger-like and pedunculated polypoid masses with vellus hair and follicular units deep within the adipose tissue (6).

Rhabdomyomatous Mesenchymal Hamartoma present as solitary (1, 2, 6, 9) or multiple (7) dome-shaped or polypoid masses in the periorbital, perioral and periauricular areas but most occur on the chin (4– 6, 11). Ashfaq *et al* proposed that the prevalence of chin lesions suggested aberrant development of the platysma muscle (10). In the index case, the lesions were small, multiple and mainly located on the central area of the face.

The vast majority occur in a cutaneous location but more recently lesions indistinguishable from the cutaneous variety have been reported from the orbit and tongue (12, 13) and these lesions are redefining the usual clinical presentation. Irrespective of location, most cases present between the neonatal period and childhood although adult presentations have also been recorded (6, 14).

The male preponderance of the first five cases raised the question of an X-linked inheritance (15) but subsequent reports in females mitigate against this genetic determination (14, 16, 17). Furthermore, increasing numbers of cases have been documented in females such that the male: female ratio in 28 reported cases to date, including this case is 1.5:1.

White suggested that the presence of RMH may indicate concomitant associated congenital abnormalities (18). These include cleft lip and cleft gum, amnion rupture sequence and Delleman's syndrome consisting of colobomas, absent corpus callosum, orbital cyst, proencephalic cysts and facial skin tags (2, 6). Nevertheless, only seven cases have had other associated congenital abnormalities. No other congenital abnormality was detected in this case.

For the most part, RMH are inconsequential lesions but may be large and occasionally capable of spontaneous movement (6, 7). Treatment involves local excision for cosmesis and no recurrence has so far been recorded. In the index case with multiple small lesions, the remaining lesions became inapparent on clinical follow-up, suggesting the possibility of spontaneous regression in small lesions. Re-biopsy was not indicated.

Although rhabdomyomatous mesenchymal hamartomas, for the most part, are only of cosmetic importance, when large and multiple they may occasionally be associated with, and therefore indicative of, other congenital abnormalities. Although they remain a rare occurrence, these entities are being recognized increasingly in females and therefore the initially suspected X-linked inheritance and male exclusivity no longer apply. Continued reporting will facilitate the elucidation of their clinical features, natural history and relationship to disordered embryogenesis.

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