Rhabdomyomatous mesenchymal hamartoma presenting as a big subcutaneous mass on the neck: a case report

YuLan Wang*, HaiHua Zhao, XinHua Yue, XinPing Tang, Li Ma, Yong Fu and PeiYi Zhang

Abstract

Introduction: We describe the location, size, histopathologic aspect and immunohistochemical expression of a rhabdomyomatous mesenchymal hamartoma, with the aim of providing useful information for its correct diagnosis.

Case presentation: A 31-year-old Chinese man first presented 2 years previously with a solitary subcutaneous mass on the left side of his neck and under his mastoid process; the mass’s size was 2×2cm. The mass increased in the size in the past 2 years. Magnetic resonance imaging revealed a dumbbell shaped and well-outlined highly reflective mass, with its upperpart infiltrating the interspace of the atlanto-occipital joint. The mass was surgically removed. On macroscopic examination, the mass was oblong and partly encapsulated, the size of the mass was 4.9×3.5×3cm, and its cut side was grey. On histologic examination, it showed a disordered collection of bundles of mature striated muscle fibres arranged in a haphazard manner and interspersed with adipose tissue, fibrocytes or mesenchymocytes and collagen, and had a myxoid matrix. On immunochemical examination, mature striated muscle was positive for desmin and myoglobin, adipose tissue and nerves were positive for S-100 protein, and fibrocytes or mesenchymocytes and collagen were positive for vimentin and cluster of differentiation 34. A diagnosis of rhabdomyomatous mesenchymal hamartoma was established.

Conclusions: Rhabdomyomatous mesenchymal hamartoma is a rare dermal or subcutaneous lesion, and we describe its immunohistochemical expression for the first time. This case report provides more information on the microscopic appearance and immunohistochemical expression

Keywords: Immunohistochemistry, Neck, Rhabdomyomatous mesenchymal hamartoma
oblong and partly encapsulated, the size of the mass was 4.9×3.5×3cm, its cut side was grey, and it felt firm. On histologic examination, it showed a disordered collection of bundles of mature striated muscle fibres arranged in a haphazard manner and interspersed with adipose tissue, fibrocytes or mesenchymocytes and collagen, and had a myxoid matrix, mitotic figures were typically rare or absent altogether (Figure 1). On immunohistochemical examination, mature striated muscle was positive for desmin and myoglobin (Figure 2), adipose tissue and nerves were positive for S-100 protein, and fibrocytes or mesenchymocytes and collagen were positive for vimentin and cluster of differentiation 34 (CD34; Figure 3). The clinical, macroscopic, histologic, and immunohistochemical characteristics allowed diagnosis of RMH.

Discussion

RMH was first described as striated muscle hamartoma in 1986 by Hendrick et al. [2], Mills [3] first used the term RMH in 1989 to describe this benign hamartoma. Since 1986, more than 30 cases of RMH have been recognized and reported in the literature. This entity exists under various names including striated muscle hamartoma, congenital midline hamartoma, and hamartoma of cutaneous adnexa and mesenchyme.

RMH is a rare dermal or subcutaneous lesion comprising disordered mature adipose tissue, skeletal muscle, adnexal elements, nerve bundles and collagen [4]. There is no evidence of cellular or malignant degeneration. RMH occurs most commonly in areas where there is superficial striated muscle, such as the nose or chin, followed by the periorbital and anterior neck areas. Most cases have been described in young patients on the head and neck [1], such as periorbital [5], nasal alae [6], lip [7], chin [8], tongue and tonsil [9], and sternoclavicular area [10]. There is no apparent sex predilection. On clinical examination, most RMH are solitary, with a few patients having multiple lesions. Most lesions present as a nodule, papule, skin tag or mass; there was a case presenting as a depressed skin lesion [11]. On macroscopic examination, the size of a lesion varied from 0.3cm to 1.4cm in the reported literature.

In our case the histology was similar to that described for RMH; however the lesion’s size was 4.9×3.5×3cm, was bigger than any other reported cases. To our knowledge, this is the biggest lesion of RMH reported. The mass was partly encapsulated. On histological examination, RMH comprises disordered mature adipose tissue, skeletal muscle, nerve bundles and collagen. The reported cases did not describe the immunohistochemical expression of RMH. In this case, skeletal muscle was positive for desmin and myoglobin, adipose tissue and nerves were positive for S-100 protein, fibrocytes or mesenchymocytes and collagen were positive for vimentin and CD34. It is a new find that the lesion’s stromata were positive for CD34.

The aetiology of RMH is unknown, possible explanations include aberrancy in the embryonic migration of mesodermally derived tissues or a genetic defect predisposing to the formation of hamartomas. Although an association with congenital abnormalities is uncommon, this possibility should be assessed by the clinician.

Figure 1 The mass was composed of disordered skeletal muscle and infiltrated the adipose tissue, fibrocytes or mesenchymocytes and collagen (hematoxylin and eosin stain, original magnification ×10). Adipose tissue and fibrocytes or mesenchymocytes were admixed with the muscle tissue in varying amounts throughout the lesion.
These lesions are benign, and are typically only excised if causing mass-type effects or for cosmetic reasons.

Conclusions
RMH is a rare dermal or subcutaneous lesion, and we describe its immunohistochemical expression for the first time. This case report provides more information on the microscopic appearance and immunohistochemical expression of RMH.

Consent
Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Level of interest
The article is important in the clinical pathology field.
Abbreviations
CD34: Cluster of differentiation 34; RMH: Rhabdomyomatous mesenchymal hamartoma.

Competing interests
The authors declare that they have no competing interests.

Authors’ contributions
YLW performed immunohistochemical assays, analysed data, and wrote the manuscript; HHZ and XHY contributed to the histological examination; XPT contributed to perform histological techniques; LM contributed to perform immunohistochemical techniques; and YF and PYZ analysed and interpreted the patient data regarding histological and immunohistochemical examinations. All authors read and approved the final manuscript.

Acknowledgements
We thank Yong Teng for supplying the patient’s clinical data. We also thank the patient described in this case report for his consent to publish the findings.

Received: 19 April 2014 Accepted: 26 October 2014
Published: 6 December 2014

References
1. Diaz-Perez JA, Garcia-Ramirez CA, Garcia-Vera JA, Melo-Urbe MA, Uribe CJ: Rhabdomyomatous mesenchymal hamartoma. Actas Dermosifiliogr 2008, 99:474.
2. Hendrick SJ, Sanchez RL, Blackwell SJ, Raimer SS: Striated muscle hamartoma: description of two cases. Pediatr Dermatol 1986, 3:153–157.
3. Mills AE: Rhabdomyomatous mesenchymal hamartoma of skin. Am J Dermatopathol 1989, 11:58–63.
4. Rosenberg AS, Kirk J, Morgan MB: Rhabdomyomatous mesenchymal hamartoma: an unusual dermal entity with a report of two cases and a review of the literature. J Cutan Pathol 2002, 29:238.
5. Ferguson JW, Hutchison HT, Rouse BM: Ocular cerebral and cutaneous malformations: confirmation of an association. Clin Gen 1984, 25:464–469.
6. Sanchez RL, Raimer SS: Clinical and histologic features of striated muscle hamartoma: possible relationship to Delleman’s syndrome (case report). J Cutan Pathol 1994, 21:40–46.
7. Dal Vechio A, Nakajima E, Pinto D Jr, Azevedo LH, Migliari DA: Rhabdomyomatous (mesenchymal) hamartoma presenting as haemangioma on the upper lip: a case report with immunohistochemical analysis and treatment with high-power lasers. Case Rep Dent 2013, 2013:943953.
8. Ashfaq R, Timmons CF: Rhabdomyomatous mesenchymal hamartoma of skin. Pediatr Pathol 1992, 12:731–735.
9. Magro G, Di Benedetto A, Sanges G, Scalisi F, Alaggio R: Rhabdomyomatous mesenchymal hamartoma of oral cavity: an unusual location for such a rare lesion. Virchows Arch 2005, 446:346–347.
10. Araceli S-C, Roberto V-G, Cirilo S-A: Rhabdomyomatous mesenchymal hamartoma presenting as a skin tag in the sternoclavicular area. Pathol Oncol Res 2007, 13:375–378.
11. Ball EAM, McGrath EJ, Chong H, Moss ALH: Rhabdomyomatous mesenchymal hamartoma resembling scleroderma ‘en coup de sabre’: a case report and literature review. Br J Dermatol 2010, 162:222–224.