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CASE REPORT

Rapidly involuting congenital hemangioma: a case report

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Abstract

Introduction: Congenital hemangioma is an uncommon benign vascular neoplasm related to mutations of the GNAQ and GNA11 genes. It develops intrauterinely and is therefore present at birth. Depending on its clinical behavior, it can be classified as rapidly involuting, non-involuting and partially involuting. **Case report:** Newborn, 25 days old, exhibiting a greenish cervical nodule on the left since birth, associated with telangiectasia. There was no growth of the lesion between birth and dermatological evaluation. A soft tissue ultrasound was performed on the first day of life and a diagnosis of hemangioma was made. **Discussion:** Rapidly involuting congenital hemangiomas usually present as single lesions, which may or may not be associated with other vascular lesions in solid organs. The diagnosis is predominantly clinical and can be complemented by ultrasound evaluation. Their regression occurs spontaneously between 6 and 14 months, for that reason expectant management is indicated in most cases.

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INTRODUCTION

Vascular anomalies in pediatrics may be congenital or arise during childhood and are classified as vascular neoplasms or vascular malformations. Most vascular anomalies are benign, but they may be associated with significant morbidity. Among vascular anomalies, infantile hemangioma stands out as the most common, and it also has a congenital variant known as congenital hemangioma (CH)[1].

CH is rare (0.3%) and shows no sex predilection[2]. It develops in utero and is present at birth. It typically presents as a solitary lesion located on the extremities, head, or neck. Three types of CH have been described: rapidly involuting congenital hemangioma (RICH), non-involuting congenital hemangioma (NICH), and partially involuting congenital hemangioma (PICH)[1].

The main characteristics of CH include a solitary, well-demarcated, erythematous-violaceous lesion with telangiectasias on the skin surface, surrounded by a pale halo. It may present as an elevated violaceous soft-tissue mass with prominent peripheral veins or as a soft-tissue mass[2].

The aim of this study is to report a case of CH, its clinical characteristics, the importance of accurate diagnosis, its possible systemic repercussions, and therapy.

CASE REPORT

A 25-day-old male newborn was referred to the pediatric dermatology outpatient clinic due to a left cervical nodular lesion present since birth. At the first dermatology visit, an oval, well-demarcated nodule with a fibroelastic consistency was observed, with a slightly greenish coloration, a pale halo around the lesion, and telangiectasias on the surface of the affected area, measuring approximately 1.8 × 1.0 cm, located in the left anterior cervical region (Figure 1).

A soft-tissue ultrasound was requested for better characterization of the cervical lesion, as well as a complete abdominal ultrasound.

The patient returned at 1 month and 14 days for a new evaluation and review of the ultrasound results. Clinically, the cervical lesion showed slight regression in size, while maintaining the characteristics observed at the first assessment. Ultrasound identified a hypoechoic lesion involving the dermis and hypodermis, composed of vessels, with well-defined margins and intense Doppler flow, suggesting hemangioma as a possible diagnosis, with vascular malformation as a differential diagnosis. The abdominal ultrasound showed no abnormalities. The main diagnostic hypothesis was congenital hemangioma, and an expectant management approach was chosen.

At 3 months and 11 days of age, the patient returned with a marked reduction of the lesion, measuring 0.8 × 0.8 cm, maintaining a slightly greenish coloration and telangiectasias on the surface. Given the significant regression, the diagnosis of congenital hemangioma was maintained, classified at that time as the rapidly involuting type.

At 5 months and 14 days, the patient returned with complete regression of the lesion, with only subtle telangiectasias remaining in the previously affected area (Figure 2).



Figure 1. Left cervical nodular lesion identified at the initial evaluation, at 25 days of life.

DISCUSSION

There is a growing body of evidence indicating that most, if not all, CHs, regardless of their involution rate (rapidly involuting, non-involuting, or partially involuting), may be attributed to somatic activating mutations in the GNAQ and GNA11 genes. This suggests that RICH, NICH, and PICH should be considered a disease spectrum rather than distinct entities[3].

Patients with RICH almost always present with a single cutaneous lesion present at birth, as in the reported case. Lesion size may range from a few centimeters to more than 10 cm. Different clinical presentations have been described in the literature: an elevated violaceous soft-tissue mass with prominent peripheral vessels; an elevated subcutaneous mass with overlying telangiectasias associated with a perilesional pale halo[4,5]. Other less frequent variants include a pinkish-



Figure 2. Patient at 5 months and 14 days of life, with complete involution of the left cervical lesion.

violaceous tumor located in the deep dermis, an exophytic vascular tumor with central ulceration, and a vascular tumor with overlying pustules[6,7].

Diagnosis is fundamentally clinical, with the lesion recognized at birth in most cases. When there is diagnostic uncertainty, soft-tissue ultrasound is the first-line examination. On ultrasound, CHs exhibit heterogeneous structures, diffuse vascularization, high vascular density, and occasionally calcifications. On Doppler evaluation, the lesion is high-flow[8,9]. Some cases may be diagnosed in utero by ultrasound in the second or third trimester of pregnancy[10].

According to the International Society for the Study of Vascular Anomalies (ISSVA), RICH is classified as a benign vascular tumor[11]. Despite its benign nature, it may be associated with hemangiomas in other organs, especially the liver, and with rare complications such as thrombocytopenia[12], coagulopathy[12], and heart failure[13,14]. Complications such as ulceration and bleeding may also occur, particularly in lesions of larger diameter. Therefore, the importance of an accurate diagnosis is emphasized.

Regression of RICH begins soon after birth, with complete regression occurring between 6 and 14 months of life[2]. This involution may result in areas of atrophy, changes in skin color and texture, alopecia, scarring, and persistent telangiectasias. In rare cases, involution may occur during the intrauterine period[15].

As it is a self-limited condition with spontaneous regression, expectant management is indicated for patients with RICH. Unlike infantile hemangioma, RICH does not have an active proliferative phase after birth; therefore, the use of beta-blockers such as propranolol is not effective. Larger lesions associated with functional impairment or concomitant hepatic lesions should be followed by a multidisciplinary team.

Identification of CH and knowledge of its behavior are essential so that appropriate diagnostic tests can be requested to confirm the diagnosis, as well as to avoid unnecessary medical interventions.

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