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Case Report

Rhabdomyomatous Mesenchymal Hamartoma Presenting as a Midline Nasal Papule in Early Infancy: Case Report

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Abstract

Hamartomas are benign, tumor-like malformations composed of an abnormal mixture of tissue elements normally found at the site of origin but arranged in a disorganized fashion. Rhabdomyomatous Mesenchymal Hamartoma (RMH) is a rare subtype, first described in 1986, characterized by a haphazard proliferation of mature skeletal muscle fibers admixed with other mesenchymal and adnexal elements within the dermis and subcutis. RMH most commonly presents as a congenital, solitary, dome-shaped papule or nodule in the midline head and neck region of infants and young children, though cases in older children and adults, as well as in atypical locations, have been reported. We present the case of a 2-month-old female with a congenital midline nasal lesion that enlarged proportionally with growth. Physical examination revealed a firm, dome-shaped papule on the inferior columella. MRI and MRA excluded intracranial extension. Surgical excision at 14 months demonstrated haphazardly arranged skeletal muscle bundles within the dermis, interspersed with hair follicles and adipocytes with desmin positivity and SMA negativity on immunohistochemistry, confirming the diagnosis of RMH. Surgical excision remains curative with no reported recurrences or malignant transformation. This case highlights the importance of considering RMH in the differential diagnosis of midline facial papules in infants and demonstrates the value of clinicopathologic correlation and imaging to guide management.

Keywords: Rhabdomyomatous Mesenchymal Hamartoma; Congenital Lesion; Nasal Midline Lesion

Abbreviations:

MRI: Magnetic Resonance Imaging; MRA: Magnetic Resonance Angiography; RMH: Rhabdomyomatous Mesenchymal Hamartoma; SMA: Smooth Muscle Actin

Introduction

Hamartomas are benign, tumor-like malformations composed of mature tissue elements native to the site of origin but arranged in a disorganized fashion; they may arise in the skin and subcutis and can mimic neoplastic processes necessitating clinicopathologic correlation for accurate diagnosis [1-3]. Unlike neoplasms, hamartomas do not exhibit true clonal expansion or malignant potential but rather represent developmental anomalies resulting from aberrant growth or differentiation during embryogenesis [1,3,4]. These lesions can arise in virtually any organ system, including the skin, subcutis and nervous system and may be composed of mesenchymal, epithelial or mixed tissue components [1-4]. Rhabdomyomatous Mesenchymal Hamartoma (RMH) is a rare, benign congenital lesion characterized by a disorganized proliferation of mature skeletal muscle fibers admixed with adipose tissue, fibrocollagenous stroma and adnexal structures within the dermis and subcutis [5-8].

RMH most frequently presents as a solitary, polypoid or nodular lesion in the midline head and neck region of infants and young children, though cases have been reported in perianal, lingual and acral locations and rare adult-onset and plaque-type variants have been described [9-11]. While most lesions are isolated and asymptomatic, RMH may be associated with congenital anomalies or syndromic features, necessitating careful clinical evaluation [11].

Recognition of the clinicopathologic spectrum of RMH is essential to avoid misdiagnosis and overtreatment, as simple surgical excision is typically curative and the prognosis is excellent [12-14]. We present a case of RMH arising in the nasal columella of an infant, with emphasis on clinical and histopathologic features.

Case Report

A 2-month-old female, born at term via vaginal delivery after an uncomplicated pregnancy, was referred to the Department of Dermatology at the University of North Carolina (UNC) for evaluation of a congenital nasal lesion. Her parents had noted a small papule on the nostril at birth, which enlarged proportionally with her facial growth. The lesion was asymptomatic, without drainage, pulsation or changes in size during crying or Valsalva maneuvers. On physical examination, a 3-mm dome-shaped, skin-colored papule was observed on the inferior columella, precisely at midline (Fig. 1). The surface was smooth and intact, with no ulceration, telangiectasia, punctum or surrounding erythema. The lesion was firm, non-tender and immobile over deeper structures. A soft tissue ultrasound in the newborn period was inconclusive. Subsequent MRI/MRA revealed no evidence of intracranial extension, sinus tract formation or associated bony defects, although evaluation was limited by the lesion's small size.

The patient was referred to UNC pediatric otolaryngology and at 14 months of age underwent surgical excision. Grossly, the lesion was a well-circumscribed nodule in the superficial dermis.

Histopathology and Immunohistochemistry

Histopathologic examination revealed epidermal acanthosis with papillomatosis and numerous skeletal muscle bundles haphazardly embedded within the superficial dermis, interspersed among benign-appearing hair follicles and fibrous stroma (Fig. 2). Rare adipocytes and adnexal structures were present. Immunohistochemistry demonstrated diffuse desmin positivity (Fig. 3) and SMA negativity, confirming the striated muscle origin. These findings, in conjunction with the lesion's clinical morphology and location, were diagnostic of RMH.

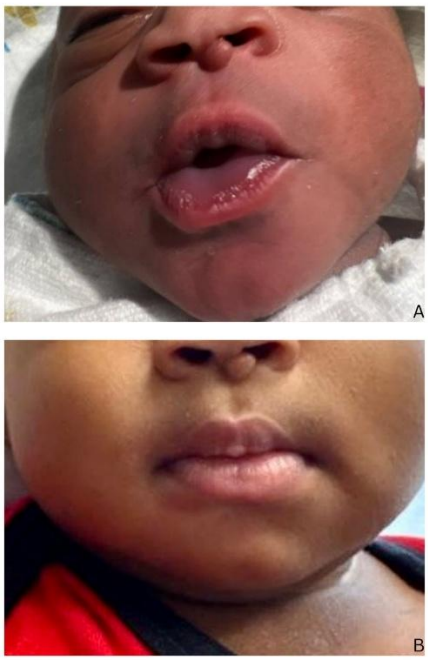

Figure 1: Clinical images showing a 3-mm dome-shape, skin-colored papule on the inferior columella. Lesion has a smooth, intact surface and is centrally located at the midline (A, B).

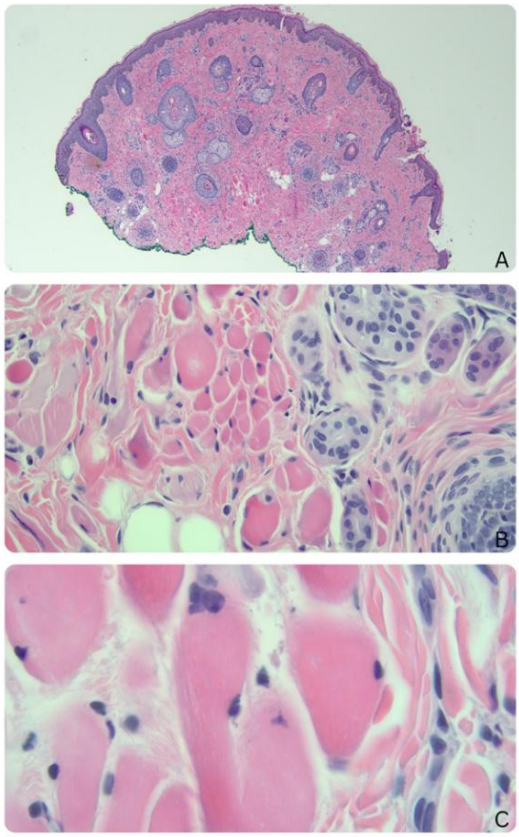

Figure 2: Histopathologic examination with hematoxylin and eosin (H&E) staining. Low power (4X, A) shows epidermal acanthosis and skeletal muscles extending into the papillary dermis. Higher magnification (40X, B; 100X, C) reveals haphazardly arranged striated muscle fibers interspersed among benign-appearing hair follicles and fibrous stroma.

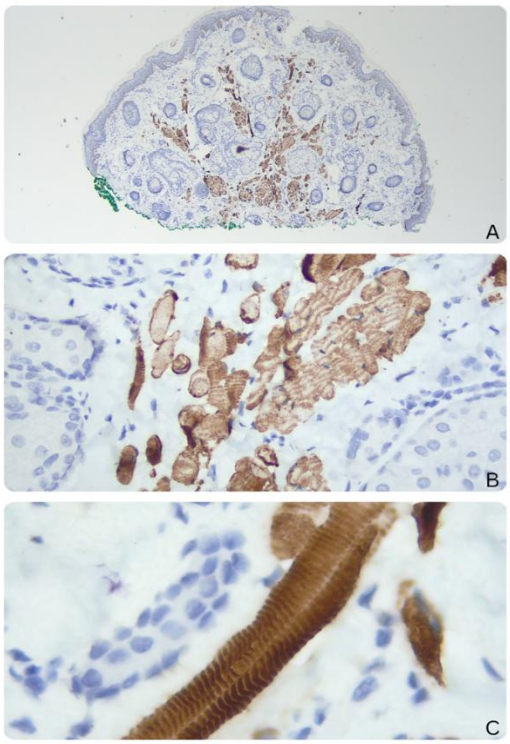

Figure 3: Immunohistochemistry for desmin confirms skeletal muscle origin. Desmin-positive staining highlights muscle bundles within the superficial dermis at low (4X, A), intermediate (40X, B) and high (100X, C) magnification.

Discussion

RMH represents a rare, benign congenital lesion characterized by a disorganized proliferation of mesodermal and ectodermal components, including mature skeletal muscle fibers, adipocytes, fibrous stroma and adnexal structures within the dermis and subcutis. It typically presents in infancy or early childhood as a solitary, asymptomatic, skin-colored papule or polyp, most frequently along the midline of the face or lumbosacral region [5]. Synonyms include striated muscle hamartoma, congenital midline hamartoma and hamartoma of cutaneous adnexa and mesenchyme [6,7]. Although RMH is most often diagnosed in neonates, later pediatric and adolescent presentations have been reported. These typically reflect congenital lesions that had not previously come to clinical attention though rare cases of acquired or trauma-associated lesions have also been described [8-11]. The pathogenesis remains unclear but may involve aberrant migration of mesodermal tissue or defective development of branchial arch derivatives during embryogenesis [12].

Histopathology

RMH demonstrates haphazardly arranged skeletal muscle fibers in the dermis and sometimes the subcutis, interspersed with mature adipocytes, fibrocollagenous stroma and adnexal structures. Immunohistochemistry typically shows desmin-positive, SMA-negative staining of muscle fibers. S100 may highlight adipocytes or neural elements, while CD34 highlights stromal components [6-8,10,11]. This admixture of tissues can pose diagnostic challenges and necessitates distinction from other mesenchymal or hamartomatous lesions.

Differential Diagnosis

Clinically, lesions arising along the nasal midline often raise concern for congenital anomalies such as nasal dermoid sinus cysts, nasal gliomas or encephaloceles [1-3,15]. These entities differ from RMH in origin, composition and behavior. Dermoid cysts are composed of ectodermal elements, lacking the mesenchymal admixture seen in RMH and may form sinus tracts with potential intracranial extension [16,17]. Nasal gliomas consist of heterotopic glial tissue and are typically firm, non-pulsatile and located laterally or paramedially [18]. Encephaloceles represent herniations of intracranial contents through skull defects and often fluctuate in size with crying or straining, unlike RMH [18]. Radiologic evaluation, including MRI and MRA, is essential in distinguishing these lesions from RMH and in excluding central nervous system involvement when midline facial lesions are present [13]. Histopathologically, the differential includes both congenital midline lesions and polypoid dermal hamartomas [6]. Epidermal nevi and fibroepithelial polyps, the latter more common in adults, lack intradermal skeletal muscle and are therefore readily distinguished from RMH. Hairy polyps can contain cartilage or glial tissue, but unlike RMH, do not demonstrate diffuse interspersion of muscle fibers. Neuromuscular hamartomas, on the other hand, typically arise in deeper locations and are closely associated with nerve trunks. Fetal rhabdomyomas may mimic RMH but are composed of immature myocytes without accompanying adnexal or adipose elements. Smooth muscle hamartomas differ immunohistochemically, showing SMA positivity and favoring the trunk and extremities [6,7]. Finally, other entities such as nevus lipomatosus superficialis and fibrous hamartoma of infancy contain ectopic adipose or fibrous/myxoid tissue, respectively, but neither exhibits the striated muscle bundles characteristic of RMH [5-7].

Clinical Course and Management

RMH is a distinct clinicopathologic entity characterized by mature skeletal muscle fibers embedded in a disorganized dermal matrix, often adjacent to other benign mesenchymal elements [5-8,16,17]. Accurate diagnosis requires correlation between clinical and histologic findings, particularly when initial superficial biopsies are nondiagnostic. In such cases, deeper tissue sampling may be necessary. Surgical excision is definitive and has not been associated with recurrence or malignant transformation. While treatment is typically not required due to the lesion's benign nature, excision may be pursued for symptomatic relief or cosmetic reasons. Rare reports of spontaneous regression have also been described [7,8,14].

Conclusion

RMH is a rare but distinct benign lesion of infancy that should be considered in the evaluation of midline facial papules. Thorough clinical examination, appropriate imaging and histopathologic confirmation are essential for diagnosis. Surgical excision is both diagnostic and curative with an excellent prognosis.

Conflicts of Interest

The authors declare no conflict of interest in this paper.

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Authors' Contributions
All authors contributed to conceptualization, treatment execution, manuscript writing and final approval.

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