

Dermatorynchus geneae: A seldom-described first branchial arch deformity

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INTRODUCTION

This case report addresses dermatorynchus geneae, a seldom-described deformity of the first branchial arch. Two previous case reports discuss this clinical entity, both of them published in the 1970s. Bittmann and Søgaaard¹ were the first to report the case of a 1-month-old girl who presented with an unusual 2-cm growth on the skin overlying the zygoma. Histologic analysis of the lesion found that it was composed of mature tissues arising from multiple embryonic germ layers, leading the authors to classify it as a branchial arch deformity. They named the as-yet undescribed entity *dermatorynchus*—literally, *skin snout*.

CASE REPORT

Our patient was a 5-week-old girl who presented with a lesion on the submental chin. This lesion had been present and enlarging since birth. The mother reported an uncomplicated pregnancy with unremarkable pre- and postnatal care. On examination, protruding from the midline submental chin, we found a 1-cm, compressible, snout-shaped lesion covered with normal skin (Fig 1). The patient did not have any facial, ophthalmic, or otic asymmetry. Ocular movements and facial expressions were appropriate for age. No other physiologic abnormalities were found on examination. Because of the lesion's anatomic location, we initially considered an unusually shaped thyroglossal duct cyst. However, the lesion did not move while the patient was feeding or swallowing. When excised, the lesion bled freely and had muscle fibers connecting it more deeply to the underlying structures of the neck.

On histologic evaluation, the lesion had a polypoid configuration and numerous vellus hairs

with affiliated sebaceous lobules. Adipose lobules were also present centrally. At the core of the lesion, striated muscle fibers were arranged both longitudinally and circumferentially. No cartilage or bone was identified (Figs 2-4).

DISCUSSION

When Bittmann and Søgaaard¹ first described dermatorynchus geneae in 1974, they presented the case of a 1-month-old girl with an elastic, snout-shaped lesion on her zygomatic cheek. The lesion was covered with normal skin. On examination, the patient was not found to have any other physical or developmental abnormalities. On removal, the lesion had a firm central cord attaching it to the zygomatic bone. On histologic examination, the lesion was shown to have a normal epidermis with associated adnexal structures (hair follicles, sebaceous glands) and an axial cord consisting of 4 arteries, 8 nerves, and longitudinal striated muscle fibers. A concentric muscular layer surrounded the longitudinal muscle fibers. No central cartilage, bone, cysts, or ducts were seen. The authors named this previously undescribed branchiogenic malformation *dermatorynchus geneae*.

In 1976, Drut and Barletta² described a similar lesion on the left cheek of a 1-month-old girl. Anterior and inferior to the left ear, the patient had a soft, 2-cm lesion covered by normal skin. On physical examination, the patient was found to have accompanying macrostomia, a component of the first branchial arch syndrome. No other physical or developmental abnormalities were noted. On removal and histologic examination, the lesion was strikingly similar to the previously reported case of dermatorynchus geneae. In addition to the pathologic features previously

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Fig 1. Lesion before biopsy.



Fig 2. Lesion histology shows polypoid lesion configuration, vellus hairs, and other normal skin appendages and subcutaneous tissues. (Hematoxylin-eosin stain. Original magnification: $\times 20$.)

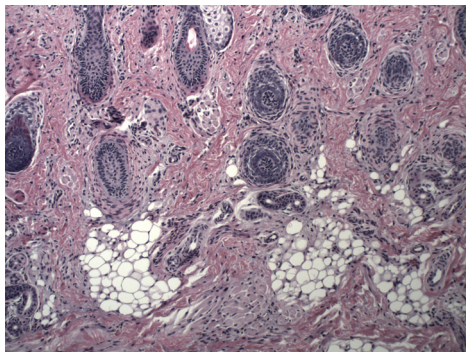


Fig 3. Lesion histology shows hair follicles, appendageal structures, subcutaneous tissue, including fat, and longitudinal and circumferentially oriented muscle fibers. (Hematoxylin-eosin stain. Original magnification: $\times 100$.)

discussed, this lesion contained a central nodule of cartilage and smaller groups of bone-forming cells. Some of these were partially calcified.²

In contrast to the two previously described cases of dermatorynchus geneae in which lesions arose on the zygomatic cheek, our patient's lesion arose on the submental chin. The midline chin is more fitting anatomic location for lesions such as a rhabdomyomatous mesenchymal hamartoma or a

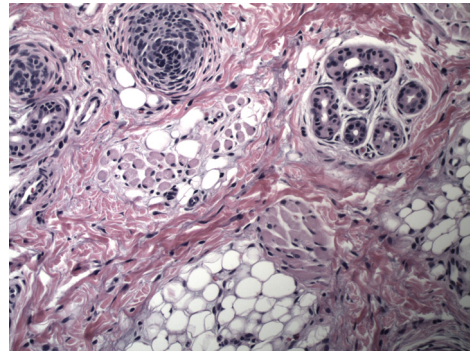


Fig 4. Lesion histology with magnified view of appendageal structures and subcutaneous tissue includes longitudinal and circumferentially oriented muscle fibers. (Hematoxylin-eosin stain. Original magnification: $\times 200$.)

wattle (cartilaginous rest in the neck).³ However, although rhabdomyomatous mesenchymal hamartomas may contain striated muscle fibers, they do not contain the circumferentially oriented muscle fibers, cartilage, or bony material typical of dermatorynchus geneae. And although cartilaginous rests may appear in similar anatomic locations to our lesion, no cartilage was identified on thorough pathologic examination of our patient's specimen. Because of the unusual composition of our patient's lesion—including skin, vellus hairs, subcutaneous tissue, and longitudinal and circumferentially oriented muscle fibers—we conclude that it represents a dermatorynchus geneae.

Drut and Barletta² felt that dermatorynchus was a distinct clinical entity, distinguished from other lesions by both its larger size and its unique longitudinal and circumferential striated muscle core.^{1,2} However, because of its reported location and pathologic composition, dermatorynchus geneae is indeed discussed in other literature as being closely related to both rhabdomyomatous mesenchymal hamartomas and accessory tragi.⁴

Accessory tragi are rarely associated with syndromes of the branchial arch.⁵ Such conditions include but are not limited to Goldenhar syndrome, also known as hemifacial microsomia (oculo-auriculo-vertebral spectrum), Online Mendelian Inheritance in Man (OMIM) 164210. Many congenital anomalies of the first and second branchial arches have been described in association with Goldenhar syndrome, including deformities of the middle and external ear, temporomandibular joint and mandible, facial musculature, and soft tissues. There also may be associated cardiac, vertebral, and central nervous system defects.⁶⁻⁸ Goldenhar syndrome is typically sporadic, but several familial cases exhibiting autosomal dominant inheritance have been described.⁹ In one recent case, a deletion

in the region of 22q11.2 led to speculation that this is a candidate gene for the syndrome.¹⁰

The paucity of case reports discussing dermatorynchus geneae makes it difficult to assess the association of these lesions with other physiologic abnormalities. Our patient represents only the third reported case of this clinical entity. It is uncertain whether the incidence of dermatorynchus geneae is truly low or whether the entity is simply underdiagnosed, for example, dismissed as a common skin tag or accessory tragus. The lack of recent reports of dermatorynchus geneae in the medical literature may also contribute to a deficiency in clinical recognition of these lesions.

Although it is not known whether a true relationship exists between the two entities, dermatorynchus geneae and accessory tragi would appear to be similar in embryologic origin, pathologic composition, and anatomic location. Therefore, given the known association between accessory tragi and congenital syndromes affecting various body systems, we believe that any patient with a suspected dermatorynchus geneae should be evaluated for craniofacial asymmetry and any other congenital anomalies via a complete and thorough physical examination.

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