Anterior Cervical Hypertrichosis: A Case Report and Review of the Literature

Bridget E. McIlwee DO,* Patrick J. Keehan, DO,**

*Dermatology Resident, OGME-3, University of North Texas Health Science Center/TCOM, Fort Worth, TX
**Faculty, Dermatology Residency, University of North Texas Health Science Center/TCOM, Fort Worth, TX; Clinical Director and Dermatologist, Premier Dermatology, Fort Worth, TX

Abstract

Anterior cervical hypertrichosis (ACH) is a rare form of localized hypertrichosis. It typically arises sporadically and is often an isolated finding. However, familial cases of ACH have been reported in association with other aberrations including skeletal abnormalities, sensory and motor neuropathies, mental retardation, and developmental delay. We present the case of a 5-year-old female with ACH in the absence of any family history of localized hypertrichosis and without any other mental or physical abnormalities.

Introduction

Unlike hirsutism, which is an excess growth of terminal hair in androgen-dependent areas such as the face, chest, or back, hypertrichosis is an increased density of hair growth in body areas that are not androgen-dependent. Hypertrichosis may occur in generalized or localized forms.1 Here, we discuss a localized form of the condition known as anterior cervical hypertrichosis, or ACH. Though localized hypertrichoses may occur sporadically and in isolation, some forms have been reported in association with significant skeletal and neurologic abnormalities. Given the potential for morbidity, a diagnosis of ACH or any other form of localized hypertrichosis warrants a complete physical exam and additional studies as indicated to rule out associated neurological and skeletal abnormalities.

Case Report

A 5-year-old white female presented to the clinic as a new patient with complaints of hair growth. Her parents noted that at approximately 3 years of age, the patient began to grow a patch of hair on the anterior neck. The hair was light brown in color and had not changed in appearance since they first noticed it. However, the hair continued to grow longer. The parents had not pursued any treatment other than trimming the hair regularly.

The patient did have eczema, but her health history was otherwise unremarkable. Given there was no family history of unusual or localized hair growth. There was no family history of neuropathies, skeletal abnormalities, or learning disabilities.

On physical exam, the patient had a 1.5 cm patch of light brown, terminal hair growing on the anterior neck, superior to the laryngeal prominence (Figures 1, 2). There was no nevus or pigment underlying the patch of hair. There were no other patches of ectopic hair growth noted on thorough physical exam. Aside from a banal eczematous plaque on the right posterior knee, the remainder of her examination — including gross skeletal, motor and sensory exams — was unremarkable. We therefore diagnosed her with anterior cervical hypertrichosis.

Discussion

Forms of localized hypertrichosis may occur congenitally or as acquired conditions. Acquired forms of localized hypertrichosis have been reported to arise after topical medications, such as corticosteroids, androgenic hormones, methoxsalen, diphenylhydantoin, and minoxidil.2,3 Localized hypertrichosis may also arise in the settings of local trauma, cutaneous hyperemia, peripheral neuropathy, chronic inflammation, or pretribial myxedema.4 Localized hypertrichosis most commonly occurs in the sacral area (“fawn tail”), but it may also occur in lumbar, thoracic, or cervical areas along posterior midline.5,6 More rarely, anterior midline cases of localized hypertrichosis have been reported. Cases of localized hypertrichosis have also been reported on the palms, soles, and elbows.7-11

Especially when these areas of localized hypertrichosis are found along the posterior midline, they are often associated with underlying defects such as spina bifida, meningocele, scoliosis, or other bony or neurologic malformations. When localized hypertrichosis is associated with underlying skeletal or neurologic abnormalities, many can be surgically corrected; and some, like diastematomyelia, can cause permanent functional damage if not corrected with due haste.12 When localized hypertrichosis occurs in an anterior distribution, it has most commonly been associated with generalized neuropathy, though other systemic associations and rare cases of underlying anterior bony deformities have also been reported.13 Both anterior and posterior localized hypertrichosis may also occur without other associated conditions.

Anterior cervical hypertrichosis (ACH) — also called ‘hairy throat’ — is a form of hypertrichosis in which there is terminal-hair growth on the anterior midline neck, superior to the laryngeal prominence. ACH may occur in a familial or sporadic fashion. There have been several reported cases of familial anterior cervical hypertrichosis.4,5,6 In one case report of a consanguineous family with three members affected by anterior cervical hypertrichosis, all three individuals also had peripheral sensory neuropathy and bilateral hallux varus, two had subclinical motor neuropathy, and one had optic nerve atrophy as well as macular degeneration.15 These associated conditions would suggest an autosomal-recessive pattern of inheritance. However, another case report of anterior cervical hypertrichosis discussed a family in which there were seven affected members, comprising three generations. The only other health problem found within the family was Turner syndrome, which would seem to suggest an autosomal-dominant pattern of inheritance.14 In yet another case report of familial anterior cervical hypertrichosis, three family members were affected. One of the family members had mild myopia, while the other two had no other medical problems. There

Figure 1

Figure 2
was no consanguinity within the family. 14 Sporadic cases of anterior cervical hypertrichosis have also been reported. 6, 16, 17 In cases of sporadic anterior cervical hypertrichosis, the majority of patients reported have had no other associated anomalies or medical conditions. 6, 17 However, some sporadic cases of ACH have been reported in association with diffuse weakness and developmental delay; 16 posterior hypertrichosis, moderate mental retardation, abnormal EEG, microcephaly, and hallux varus; 18 and posterior hypertrichosis, moderate mental retardation, dysmorphic facies, and hyperopia.19

In early 2015, Megna et al. assessed the 40 cases of ACH that have been published to date. Twenty-seven of the published cases were familial (67.5%), and 13 were sporadic (32.5%). Literature review reveals that females are more often affected with ACH than males (75% of reported cases). In the vast majority of cases, ACH presents as a solitary disorder in women (67.5% of published cases). Twenty-four cases were familial (60%), and 13 were sporadic (32.5%). Literature review reveals that females are more often affected with ACH than males (75% of reported cases). In the vast majority of cases, ACH presents as a solitary disorder in women (67.5% of published cases).

The pattern of inheritance of ACH, the etiology of the disorder remains unknown. In contrast to lumbar hypertrichosis, in which the localized increased hair density may signal an underlying skeletal abnormality (spina bifida), ACH has yet to be reported in association with defects of underlying structures. Therefore, it is unlikely that ACH arises as a secondary effect of underlying skeletal or other abnormalities.19 Furthermore, in contrast to generalized hypertrichoses (e.g., Ambras syndrome), which some have suggested to be an atavism, ACH and other localized forms of hypertrichosis seem more compatible with a homeotic gene alteration resulting in ectopic terminal-hair growth.20, 21 However, this phenotypic change has not been substantiated in mouse models of Hox gene alterations.19

In the absence of associated abnormalities, ACH is primarily of cosmetic concern to the patients and their families. Treatments are those commonly utilized to treat unwanted hair on other areas of the body, including trimming, waxing, bleaching, electrolysis, and IPL.22-24

Conclusion

It is thought that ACH is vastly underreported. Our report represents a sporadic case of ACH in a patient without any other systemic disorders, but we wish to publish it to raise awareness of ACH and its most common associations, some of which are serious and may be treated effectively if recognized promptly.

References

Correspondence: Bridget E. McIlwee, DO, University of North Texas Health Science Center, 855 Montgomery Street, 5th Floor, Fort Worth, TX 76107; Ph: 817-735-2922; F: 817-735-5022; bridget.mcilwee@unthsc.edu

The authors have no relevant financial disclosures.