Case Report

Adolescent Vulvar Angiokeratoma Associated with Lichen Sclerosus

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A B S T R A C T

Background: We present an adolescent with multiple vulvar angiokeratomas within a background of lichen sclerosus.
Case: A 13-year-old girl presented with vulvar pruritus and wart-like vulvar lesions. Four lesions were resected because of discomfort and uncertainty of the diagnosis. Pathology revealed angiokeratomas with chronic inflammation suggestive of lichen sclerosus. Postoperatively, pruritus continued in the largest excised lesion, which was associated with lichen sclerosus, and symptoms were treated successfully with topical steroids.
Summary and Conclusion: Vulvar angiokeratomas are asymptomatic red papular lesions and are rare in the female adolescent population. In this case, the pathology revealed the rare co-occurrence of angiokeratomas and lichen sclerosus. Biopsies of vulvar vascular lesions in symptomatic adolescents are recommended. Vulvar angiokeratomas might manifest rare genetic disease in otherwise asymptomatic female patients and warrant further follow-up.
Key Words: Adolescence, Angiokeratoma, Lichen sclerosus, Anderson-Fabry disease, Lysosomal storage disease

Introduction

A wide variety of lesions occur on the vulva and whereas many are limited to the vulva, others might occur on other skin or mucocutaneous surfaces or be a manifestation of a systemic disorder.

Vulvar lesions are relatively common in adolescents and can cause a diagnostic dilemma because of the extensive differential diagnosis. Often lesions are categorized according to color and morphology to aid in the assessment. Vulvar angiokeratomas are benign superficial vascular lesions consisting of superficial vascular ectasia and hyperkeratosis, which present as red-purple to black nodules, that might be single or multiple and arise along superficial vessels.[1] The 2 most common types are solitary papular angiokeratoma and angiokeratoma of the vulva.[1] They are usually diagnosed in women ages 20-50 years and are uncommon in adolescents. The differential diagnosis is broad and includes the more common cherry hemangiomas as well as follicular abnormalities (Table 1). If lesions are asymptomatic, they can be followed expectantly. If they are symptomatic or if the diagnosis is uncertain, biopsies can be done.

We present a case of an adolescent with symptomatic vulvar angiokeratomas within a background of lichen sclerosus, an unusual case of 2 concomitant skin conditions.

Case

A healthy 13-year-old girl presented with a 1-year history of mild vulvar pruritus, increasing over time, and a 4- to 5-month history of enlarging wart-like vulvar lesions. There was no associated erythema, whitening of the skin, erosions, ulceration, crusting, or bleeding. She had no other symptoms. She denied trauma, sexual activity, or abuse. The patient denied personal or family history of genital or extragenital lichen sclerosus. She denied personal neurologic or ophthalmic symptoms or a family history of metabolic disorders.

Her general physical examination was unremarkable. Breasts were symmetric Tanner 4. Examination of external genitalia showed Tanner 4 pubic hair, normal labia majora and minora, normal external meatus and clitoris, and annular hymen. Inspection revealed several nontender, papular, red-violaceous vulvar lesions, ranging from 1 to 9 mm on the labia majus (Fig. 1). Because the pruritus was thought to be associated with the lesions and the diagnosis was unclear, surgical intervention was decided upon through shared decision-making with the patient and her family. There were 5 lesions in total. The patient and parents desired removal of all 5 lesions because of esthetic concerns and interference with personal hygiene. Four lesions were excised, and one 1-mm lesion was ablated. Pathology revealed angiokeratomas in all 4 excised lesions as well as focal subepithelial hyalinization and chronic inflammation suggestive of lichen sclerosus in the largest 9-mm lesion (Fig. 2).

Postoperatively, the excision sites were well healed after 4 weeks. The pruritus continued in the area of the largest excised lesion, which was associated with lichen sclerosus. Very mild hypopigmentation was noted at that site postoperatively. She was treated for symptom relief with triamcinolone acetonide 0.1% ointment daily for 4 weeks, then every other day for 2 weeks, then stopped. Since then she has not reported any new lesions or any further

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Table 1
Vulvar Lesions of the Female External Genitalia

Common Lesions
Angiokeratomas
Atopic dermatitis (eczema)
Allergic contact dermatitis
Cherry hemangiomas
Candidiasis
Epidermoid cysts (inflamed)
Folliculitis and keratosis pilaris
Furunculosis
Irritant contact dermatitis
Molluscum contagiosum
Vestibular papillomatosis
Uncommon lesions
Crohn's disease
Human papillomavirus infection
Hidradenitis suppurativa
Psoriasis
Urethral caruncle
Urethral prolapse
Very rare
Extramammary Paget disease
Squamous cell carcinoma
Vulvar intraepithelial neoplasia

pruritus. On physical exam, 10 months after surgery, she had no evidence of changes consistent with lichen sclerosus or any new lesions. The patient and her family were counselled about the link between multiple angiokeratomas and Anderson-Fabry disease, a rare lysosomal storage disorder, and they chose to undergo genetics testing. The patient underwent α-galactosidase testing as well as Fabry disease full gene analysis through the genetics lab. Both tests came back normal. Surveillance of her vulva is ongoing.

Summary and Conclusion

Vulvar angiokeratomas are rare in the female adolescent population and have only occasionally been described in association with lichen sclerosus.[2,3] The angiokeratomas are usually asymptomatic and can be managed expectantly. If lesions present with pain, discomfort, itching, or bleeding, they should be resected or biopsied to confirm the diagnosis. Desire to achieve definitive pathologic diagnosis by patient and/or provider, cosmetic concerns, and suspicion for melanoma in darkly colored or thrombosed lesions might also necessitate biopsy or excision.

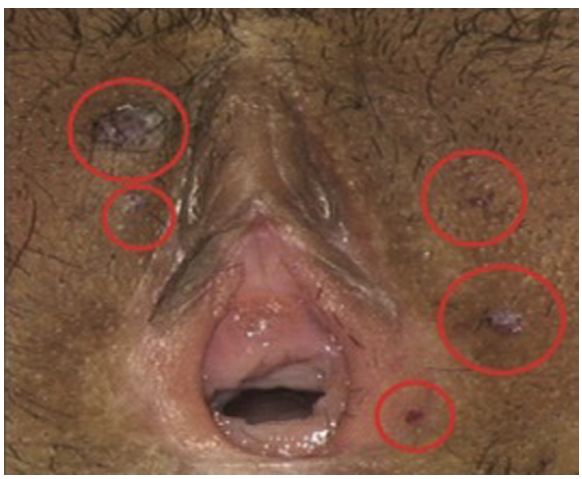

Fig. 1. External genital exam: multiple vulvar angiokeratomas are indicated by the red circles.

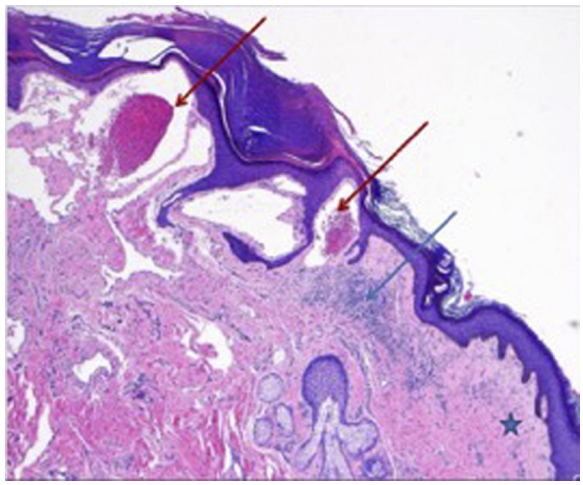

Fig. 2. Dilated subepithelial vessels consistent with angiokeratoma (red arrows), subepithelial hyalinization (star), and chronic inflammation (blue arrow), consistent with lichen sclerosus.

Angiokeratoma might be associated with Anderson-Fabry disease, which is an X-linked recessive mutation of the galactosidase alpha gene that codes for α-galactosidase A. Deficiency of the enzyme leads to progressive accumulation of globotriaosylceramide (GL-3 or Gb-3) and related glycosphingolipids in the plasma and tissue lysosomes throughout the body. This condition affects 1:40,000 to 1:60,000 men. The prevalence of Anderson-Fabry disease carriers has been reported at 1 in 339,000 women in the United Kingdom.[4] In general, affected girls have an attenuated form of the disease, with later onset and milder symptoms. In 60 carrier women neuropathic pain was the most common symptom reported by 42 women (70%), followed by fatigue that was reported by 40 women (66%), whereas angiokeratomas were present in 21 women (35%).[4] Manifestations of the disease might start in childhood, although the mean delay between the onset of symptoms and correct diagnosis of Fabry-Anderson disease has been reported at approximately 4-5 years in children and 10-14 years in adults, respectively, in 2 time periods (2001-2006 vs 2007-2013).[5] Angiokeratomas initially appear at age 5-13 years with an increase in number of lesions with age because of ongoing accumulation of glycosphingolipids.[6] Multiple vulvar angiokeratomas might necessitate a genetic workup to detect obligate female carriers who might have only cutaneous evidence of this rare lysosomal storage disease. Carrier detection with the α-galactosidase A assay is not reliable because some obligate heterozygotes have normal α-galactosidase A activity. Women at risk for carrying the disease gene should have molecular studies to detect the family's mutation. The clinical manifestations of heterozygous women range from asymptomatic with normal life span to as severe as affected men. Now that there is

enzyme replacement therapy available, with regard to microvascular endothelial deposits of globotriaosylceramide and improvement of pain-related quality of life, genetic testing has become more prevalent.[7]

Lichen sclerosus is a chronic inflammatory dermatosis which usually presents with significant itching and white parchment-like skin changes around the vagina and the anus. It can also manifest as vascular lesions, usually described as purpura. Prevalence of lichen sclerosus in young girls is reportedly 1 in 1100.[8] There is an association between lichen sclerosus and other autoimmune disorders such as autoimmune thyroiditis, vitiligo, alopecia areata, celiac disease, or type 1 diabetes. There are very few cases described in the literature of extragenital or genital lichen sclerosus associated with angiokeratomas.[2,3] This patient did not have the typical parchment appearance of the vulvar skin; however, the itching was likely because of the lichen sclerosus so the pathologic diagnosis was helpful in planning further symptomatic treatment for this patient.

This case report highlights the possibility of 2 vulvar conditions necessitating different approaches to treatment, as well as the link of vulvar disease to a rare genetic condition. Awareness by providers who care for young women might improve the diagnostic approach and subsequent treatment.

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