Teledermatology leading to an important diagnosis in an underserved clinic

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Abstract

Cutaneous signs can be the first manifestation of important medical diagnoses, including inherited cancer syndromes, but access to dermatologic evaluation is especially challenging for uninsured patients. Herein, we present a case in which a volunteer academic teledermatology triage program was used by a community health clinic to make a diagnosis of multiple cutaneous leiomyomas, which confer a high likelihood of hereditary leiomyomatosis and renal cell cancer syndrome, also known as Reed syndrome; this prompted malignancy screening for the patient. Importantly, this case underscores the potential for teledermatology to improve access to dermatologist evaluation and make crucial diagnoses in patients with barriers to care.

Keywords: teledermatology, access to care, genodermatosis, genetic cancer syndrome, cutaneous leiomyomas

Introduction

Cutaneous signs can be the initial manifestation of important and even life-threatening diagnoses [1], but access to dermatologists can be challenging for disadvantaged populations [2, 3]. We present a case in which store-and-forward teledermatology triage was used by a non-profit community health clinic to expedite an important diagnosis in an uninsured patient.

Case Synopsis

An uninsured man in his forties with no significant medical history presented to a community health clinic owing to painful lesions on the arm that first appeared four years ago. He denied noting similar lesions in family members. Given his lack of access to a dermatologist, the primary care provider submitted a photograph (**Figure 1**) for remote consultation via a volunteer-run academic teledermatology program utilizing the American Academy of Dermatology's AccessDerm smartphone application. This application provides a store-and-forward teledermatology platform to facilitate community outreach and improve access to dermatologic care [4]. The photograph demonstrated a cluster of pink papules, some pedunculated, varying from 2-6 millimeters in size. Per the examiner, the lesions were soft, tender to palpation, and located only on the right upper arm.

The differential diagnosis based on morphology and distribution included leiomyomas, neurofibromas, sclerotic fibromas, or collagenomas, all of which can be associated with genetic disorders [1]. Based on concern for an underlying disease manifesting with cutaneous findings, the teledermatologist recommended in-person evaluation and tissue sampling for histology. Biopsy of three lesions was

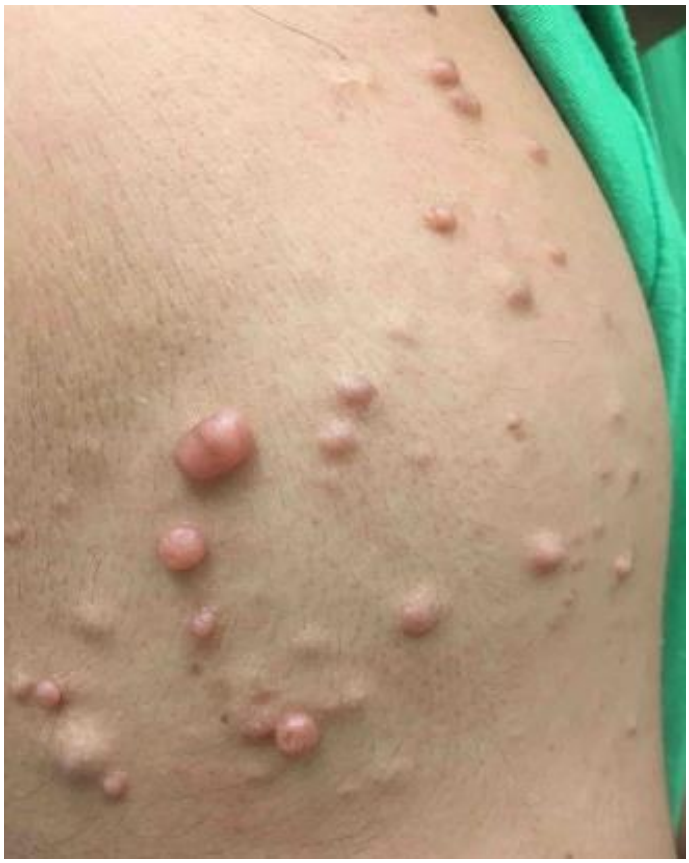

Figure 1. *Clinical photo-graph: physical exam revealed a cluster of about 30 pink tender papules, some pedunculated, on the right upper arm.*

performed at the community health clinic during the monthly volunteer dermatology clinic, which provides in-person follow-up appointments as needed for patients that are first triaged via teledermatology [5]. Histopathology revealed proliferation of mature spindle-shaped cells in the dermis with "cigar-shaped" nuclei and peri-nuclear clearing characteristic of leiomyomas (**Figure 2**).

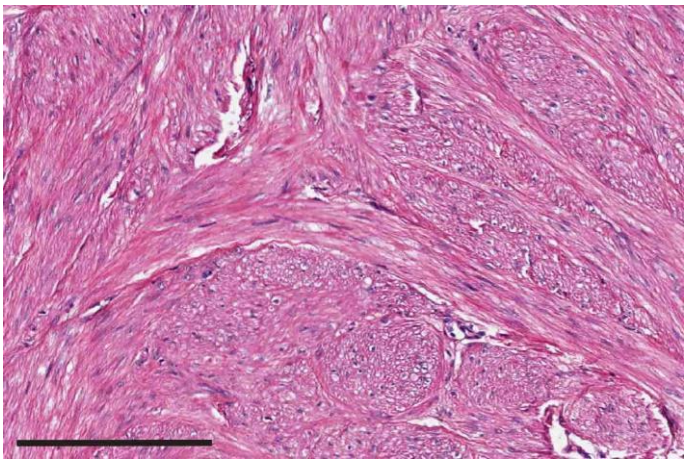

Figure 2. *Skin biopsy: shave biopsy of the lesions revealed interlacing fascicles of benign-appearing spindle-shaped cells with abundant pink cytoplasm, blunt-ended nuclei, and peri-nuclear vacuolization (H&E, 40×, scale bar = 100µm).*

Case Discussion

Importantly, histologic confirmation of multiple cutaneous leiomyomas satisfies the major criterion and confers a high likelihood of hereditary leiomyomatosis and renal cell cancer (HLRCC) syndrome (Reed syndrome), a rare inherited disease characterized by multiple benign smooth muscle tumors, most commonly cutaneous leiomyomas and uterine fibroids in women [6, 7]. For female patients, annual gynecologic exam is recommended since large, multiple, symptomatic uterine fibroids often develop at an early age and necessitate hysterectomy [7- 9]. Unfortunately, the disorder also carries an increased risk of aggressive renal cell carcinoma (RCC) with an estimated lifetime frequency of 15% [10].

HLRCC syndrome-associated RCC is particularly aggressive with the majority of patients deceased within five years of diagnosis [10], underscoring the importance of malignancy screening. Annual contrast-enhanced renal magnetic resonance imaging (MRI) remains the standard for surveillance in HLRCC [6, 10]. Computed tomography is not preferred owing to cumulative radiation exposure, and ultrasound may miss small or isoechoic tumors. For RCC in HLRCC syndrome, early aggressive surgery is advised even for small tumors [6]. In our case, the patient opted for a more affordable renal ultrasound, which showed no abnormalities, he was also referred to the clinic social worker to investigate insurance eligibility to reduce the cost of an MRI.

HLRCC syndrome has been linked to mutation of the gene encoding fumarate hydratase, a Krebs cycle enzyme [8], it is usually autosomal dominant. Thus, genetic testing provides a definitive diagnosis and should be offered to patients with suspected HLRCC syndrome and their first-degree relatives to determine whether they should undergo annual renal surveillance, which should start at 8-10 years of age [6]. Our patient was informed of his potential risk

for malignancy and the options for confirmatory genetic testing, which he declined for financial reasons.

In a large series of families with HLRCC syndrome, cutaneous leiomyomas developed by an average age of 25 years, uterine leiomyomas by 30, and renal tumors by 44 [8, 10]. Thus, cutaneous findings are usually the earliest potential indicator of HLRCC syndrome, emphasizing the importance of recognizing skin lesions to allow early diagnosis and initiation of effective malignancy screening. Typical cutaneous leiomyomas are smooth, skin-colored or pink-brown papules, often clustered and involving the extremities, trunk, or neck. Lesional pain is common and can be provoked by stroking. Treatment options for the cutaneous leiomyomas include excision, ablative laser, or intralesional corticosteroids, but symptoms may be managed with calcium channel blockers, gabapentin, or injected botulinum toxin [6].

This case underscores the potential for teledermatology to facilitate important diagnoses with cutaneous manifestations and to function as a triage mechanism for clinics serving patients lacking insurance, which is a known barrier to dermatologic care [2, 3]. To improve access to dermatologist evaluation, we established an outreach program, as utilized in this case, to combine teledermatology and an in-person clinic, both staffed by board-certified dermatologists on a voluntary basis without charge to patients. When faced with patients having cutaneous concerns that they felt unable to confidently diagnose or treat, the primary care providers were trained to utilize the American Academy of Dermatology's AccessDerm smartphone app to submit a consult request consisting of a brief clinical history and photographs taken with a smartphone camera.

Our team of volunteer teledermatologists triage the consults to determine whether a treatment plan can be established based on the history and photographs alone. If not, as for complex diseases, lesions suspicious for malignancy, or issues requiring a procedure, the patient is referred to the monthly clinic staffed by these same volunteers, thus ensuring adequate follow-up care. A recently published retrospective study reviewing this teledermatology triage system demonstrated that approximately 70% of cases were deemed manageable without the need for in-person evaluation, leading to improved availability of in-person appointments and significant reduction in the time to dermatologist evaluation, and treatment recommendations for the patients [5].

Conclusion

Regrettably, non-citizen, uninsured, and impoverished patients often lack access to specialists like dermatologists [2, 3]. However, various volunteer outreach efforts have demonstrated that telemedicine can connect remote specialist physicians to providers caring for underserved and uninsured populations [4, 5, 11-13]. In this case, teledermatology led to an important diagnosis associated with an inherited cancer syndrome and highlights the potential of telemedicine to reduce barriers to dermatologic care among disadvantaged populations.

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