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Hypohidrotic Ectodermal Dysplasia Milia Treatment With Fractional Carbon Dioxide Laser and Laser-Assisted Drug Delivery of Triamcinolone

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ABSTRACT

Hypohidrotic ectodermal dysplasia (HED) is a genetic disorder characterized by hypohidrosis, hypodontia, and hypotrichosis. Skin manifestations, including dyspigmentation and milia-like papules that coalesce into plaques, are difficult to treat. There is no cure for HED, therefore treatment is focused on managing symptoms and improving quality of life. There is limited evidence in the literature for safe and effective treatments improving HED-related facial skin aesthetics. The facial skin rashes caused by HED demonstrate an unmet clinical need in dermatology. Current therapies are limited to prevention methods such as keeping the skin cool by avoiding heat and applying topical moisturizers to help treat dry, pruritic skin. Herein we present a method for successful treatment of a 34-year-old African American male using fractional Carbon Dioxide (CO_2) ablative laser with laser-assisted drug delivery of triamcinolone 0.1% ointment that resulted in decreased milia-like papules, improved dyspigmentation, smoother skin tone, and high patient satisfaction.

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INTRODUCTION

Hypohidrotic ectodermal dysplasia (HED) is a genetic disorder caused by mutations in one or more genes involved in embryonic ectodermal development that lead to hypohidrosis, hypodontia, and hypotrichosis.[1] This disorder results from mutations in tumor necrosis factor alpha (TNF-α) and ectodysplasin A (EDA) pathways.[1] HED effects approximately 1 in 17,000 births and is most often X-linked, thus prevalence is higher in males.[1] There is no cure for HED, therefore treatment is focused on managing symptoms and improving quality of life.

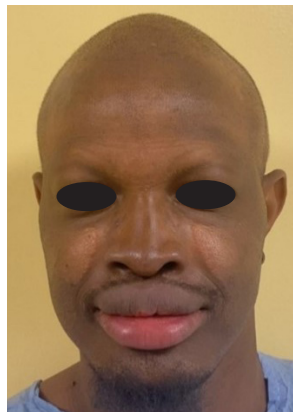
Skin manifestations of HED can be particularly difficult to treat. Symptoms include dryness, ichthyosis, hypo- or hyperpigmentation, sparse and patchy hair, and milia-like papules that coalesce into plaques on the face.[1] Milia-like eruptions in HED may be attributed to hypohidrosis, large sebaceous glands, or eruptive vellus hair cysts.[2] Treatments are limited to keeping the skin cool by avoiding heat and applying topical moisturizers to help treat dry, pruritic skin.[3]

Clinical Features of a Hypohidrotic Ectodermal Dysplasia Patient

A 34-year-old African American male presented with a persistent rash on his face that began in childhood and had not changed. The rash was not painful, burning, or pruritic, but the patient complained of significant psychological distress due to the appearance. Physical exam revealed many monomorphic milia-like papules coalescing into plaques on the face with sparing of the perioral and periorbital regions (Figure 1). Additionally, there was sparse eyebrow and scalp hair and a prominent forehead. The patient declined a full-body skin exam but had no other dermatologic concerns and no other lesions were noted in the surrounding, observable areas.

Medical history was significant for HED and multiple dental implants. The patient used adapalene gel on the face nightly

FIGURE 1. HED patient before treatment with many milia-like hypopigmented tiny monomorphic papules and coalescing plaques on the face with sparing of the perioral and periorbital regions.



but had not noticed any changes or improvement. The patient denied any other medications, allergies, family history of skin cancer, or other significant history.

Biopsy of the facial lesion at the initial visit revealed perivascular and focally perifollicular lymphocytic infiltrate with small mounds of parakeratosis. Given the patient's history of HED, the facial rash was diagnosed as HED milia-like papules that coalesce into plaques. The patient was offered fractional CO_2 ablative laser (DEKA SmartXide DOT, Calenzano (FI), Italy) with laser-assisted drug delivery (LADD) of triamcinolone 0.1% ointment. After discussion of risks, benefits, and alternatives to treatment, the patient agreed to complete three laser treatments at six-week intervals. Gentle skin care was recommended, including use of daily sunscreen and discontinuation of adapalene.

Description of Treatment Method

The patient was instructed to bring the following prescriptions to each treatment visit: topical lidocaine 2.5% and prilocaine 2.5% cream, mupirocin ointment, and triamcinolone 0.1% ointment. The patient was also given a course of 1 gram oral valacyclovir twice daily for 10 days starting one day prior to each procedure, oral cephalexin 500 mg four times daily for two weeks starting on the day of the procedure, and applied mupirocin topical ointment three times daily to the treated area for three days.

At each visit, topical lidocaine 2.5% and prilocaine 2.5% cream was applied to the treatment area including bilateral cheeks, chin, and upper cutaneous lip under occlusion for one hour prior to procedure. The facial skin was then thoroughly cleansed and de-greased with alcohol pads. Proper laser protective eyewear was worn by the patient and providers.

The CO_2 laser treatments were performed with the following settings: power 15 W, DOT mode, spacing 500 µm, repeat 2.0 seconds, Stack 1, scanning dwell 500 µs, hexagon shape, size 100%, and ratio 10/10. One pass of laser treatment with feathering to smooth the edges was administered at each of the three total treatment visits. After each session, triamcinolone 0.1% ointment was applied to the treated areas. Post-operative care was reviewed after each procedure and the patient was instructed to avoid sun.

The patient tolerated the procedures well with no complications. The patient reported mild discomfort during the procedure that did not warrant discontinuation of treatment and no serious adverse events. At the two-week follow-up visit following the final treatment, physical exam revealed decreased milia-like papules and coalescing plaques, improved dyspigmentation, and smoother, flattened skin tone (Figure 2). The patient expressed high satisfaction with the results and improved quality of life.

FIGURE 2. HED patient after fractional CO_2 ablative laser treatments demonstrating improved appearance with decreased milia-like papules, improved dyspigmentation, and smoother, flattened skin tone.

DISCUSSION

We present successful treatment of HED associated milia-like papules and coalescing plaques using a combination approach with fractional CO_2 ablative laser and LADD of triamcinolone 0.1% ointment. There is limited evidence in the literature for safe and effective treatments improving HED-related facial skin aesthetics. One case report of a 27-year-old Chinese male patient documented the use of fractional CO_2 laser treatment.[4] The patient received three laser treatments for hyperplastic sebaceous glands at six month intervals and reported marked improvement in facial skin aesthetic appearance.[4] Our method is differentiated from this documented case as we used LADD to enhance the aesthetic outcome and minimize post-inflammatory pigmentary alteration (PIPA) from CO_2 laser. Our literature search did not reveal any additional reports of laser treatment for HED. The facial skin rashes caused by HED demonstrate an unmet clinical need in dermatology.

Fractional CO_2 ablative lasers emit wavelengths of 10,600 nm with water as the target chromophore.[5] This treatment is often used for skin rejuvenation and resurfacing to improve skin texture, tone, and rhytids and to treat scars and photodamage.[5] CO_2 lasers are ablative lasers that vaporize portions of the epidermis and allow for new skin to form in its place.[6] These effects may help improve the appearance of sebaceous hyperplasia, miliaria, and dyspigmentation in patients with HED.

Patients should be counseled on the risks of CO_2 laser before beginning treatment, including dyspigmentation, burning, pain, erythema, scarring, infection, and potential outbreaks for patients with herpes simplex virus (HSV).[5,7,8] We recommend performing test spots and the use of conservative laser parameters to help mitigate side effects such as dyspigmentation. To help enhance procedure-related comfort, we advise a one-hour incubation of topical lidocaine 2.5% and prilocaine 2.5% cream under occlusion. Additional local anesthetic injections can be administered based

on patient preference. To reduce the risk of infection, we suggest patients take a two-week course of oral antibiotics following each treatment and applied topical mupirocin to the treated area three times daily for three days. To prevent HSV outbreaks, we suggest prophylactic oral valacyclovir 1 gram twice daily for 10 days starting the day before each procedure. We also advise application of triamcinolone 0.1% ointment to the treated area immediately following the procedure and sun avoidance to promote healing and to minimize PIPA and photodamage. It is important to set patient expectations that the pigmentary changes associated with the milia-may not fully resolve.

Skin of color patients may be particularly sensitive to side effects from laser treatments, including erythema, blistering, and PIPA.[9] Dermatologists may prefer to begin their skin of color patients on conservative laser parameters to determine their patient's response before gradually increasing dosing, power, and frequency.

We present a new treatment method for addressing the aesthetic concerns of HED patients using fractional CO_2 ablative laser with LADD triamcinolone 0.1%. We advise approximately three treatments spaced at a minimum of six weeks apart to allow time for the skin to heal between sessions and to prevent scarring. We suggest following up with patients one week after each treatment to monitor progress and address any adverse events. Patients may require additional sessions to achieve or maintain their desired results.

This combination approach equips dermatologists with a practical treatment for their HED patients. This treatment can be administered safely with minimal risk of side effects and serious adverse events. This case demonstrates that fractional CO_2 ablative laser with LADD triamcinolone 0.1% can be effective at improving the milia facial skin rash caused by HED that has previously been difficult to treat, while also improving HED patients' self-esteem and quality of life.

DISCLOSURES

The authors have no conflicts of interest to declare.

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