

## Case Presentation

### Successful treatment of facial papules with electrodesiccation in a patient with birt-hogg-dubé syndrome

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## Case synopsis

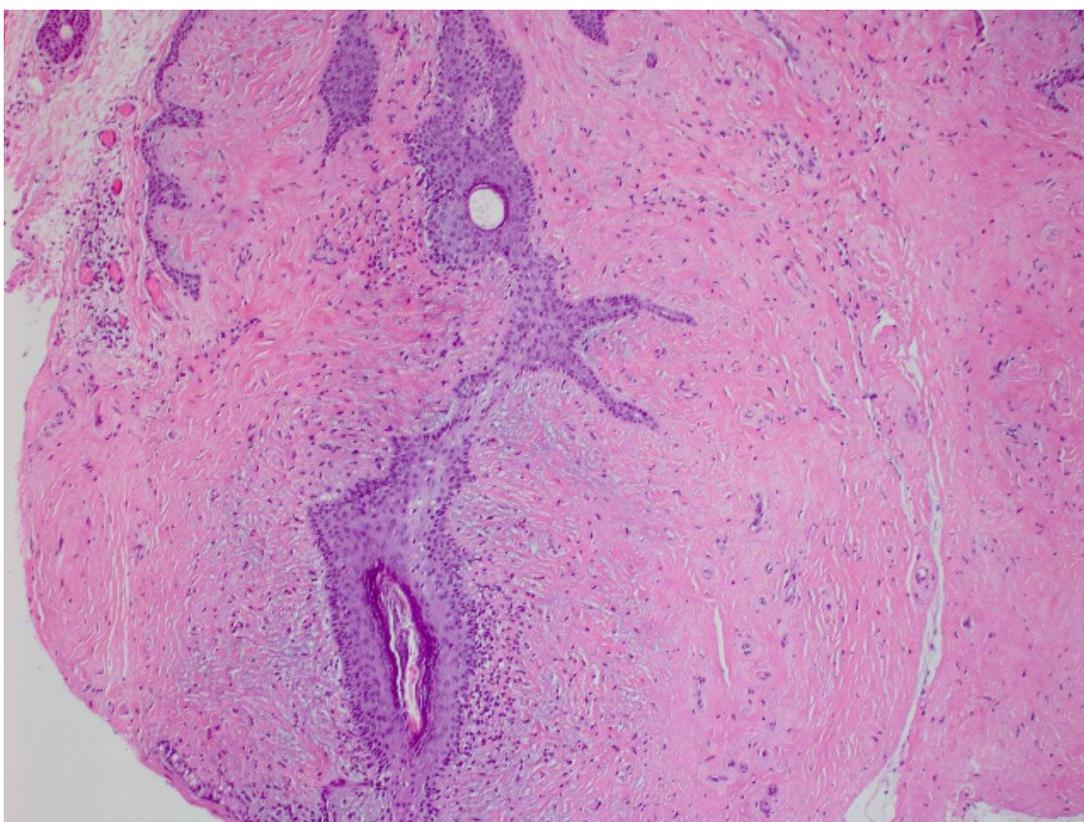
We report a case of a 51-year-old Hispanic woman who presented with a several year history of multiple flesh colored papules of cosmetic concern on the nose and medial cheeks (Figure 1). Biopsies revealed fibrofolliculoma and trichodiscoma. The patient was referred for genetic testing and was found to be positive for the FLCN gene defect, confirming a diagnosis of Birt-Hogg-Dubé syndrome. Further work-up with screening renal ultrasound and CT scan of the thorax and abdomen was unrevealing. For treatment of these skin lesions, dermasanding was attempted initially with only minimal benefit. She subsequently had multiple lesions treated with hyfrecation (also known as electrodesiccation or cautery) at a low setting and was very pleased with the results (Figure 2). Importantly, there has yet to be a recurrence of lesions two years status-post hyfrecation.



Figure 1. Before



**Figure 2.** After



**Figure 3.** Fibrofolliculoma. Depicted above is a central distorted hair follicle with surrounding basophilic, fibrous stroma

## **Discussion**

Birt-Hogg-Dubé syndrome was first described in 1977 as a syndrome involving facial papules, pulmonary cysts, and renal tumors.<sup>1</sup> The incidence of BHD is unknown, and likely under-diagnosed due to the wide variability of expression. BHD patients have a loss of function of the folliculin protein [1], the function of which is still unknown. The suggested diagnostic criteria for Birt-Hogg-Dubé syndrome consists of fulfilling one major criterion or two minor criteria. The major criteria are either 1) Five or

more adult onset fibrofolliculomas or trichodiscomas with at least one confirmed histologically or 2) Pathogenic germline mutation of FLCN. The minor criteria are 1) Multiple pulmonary cysts without an apparent cause (with or without spontaneous pneumothorax), 2) Renal cancer with early onset (<50 years old), and 3) First degree relative with BHD syndrome [2].

Often the skin manifestations are what prompt physicians to investigate for genetic mutations of the folliculin gene, as was the case in our patient. The skin lesions usually do not appear until after the age of twenty [2]. Patients typically present with numerous fleshy papules a few millimeters in diameter located on the nose and cheeks, but the papules can also be located on the forehead, neck, and upper trunk [2]. These papules are typically fibrofolliculomas, trichodiscomas, and/or acrochordons.

Biopsies of our patient revealed fibrofolliculoma and trichodiscoma. The patient was also referred for genetic testing and was found to be positive for the FLCN gene defect, confirming a diagnosis of Birt-Hogg-Dubé syndrome. Further work-up with screening renal ultrasound and CT scan of the thorax and abdomen was unrevealing. Her main concern, however, was the cosmetic appearance of her facial papules. Unfortunately, despite a variety of attempted treatments for the multiple fibrofolliculomas and trichodiscomas observed in BHD, a gold standard for treatment of these skin lesions has not been elucidated. Laser ablation, curettage plus cautery, and shave plus cautery have been reported previously in literature. Jacob et. al reports promising results for a patient after treatment with carbon dioxide and Erbium:YAG lasers for at least 8 weeks after treatment [3]. However, there has also been evidence of recurrence of facial papules associated with BHD roughly 6 months after treatment with Er:YAG laser reported in another patient [4]. Curettage plus cautery has had promising results for one patient, with only minor recurrence three years after treatment [5]. Shave plus cautery has also been reported to have removed facial lesions successfully in another single case, but there is no mention of patient follow-up [6].

Originally our patient underwent dermasanding for treatment of her facial lesions, but we saw only minimal benefit. Unfortunately, the local anesthesia and physical pressure required for this procedure can be distressing for the patient. Later, we attempted to treat with hyfrecation alone (without anesthesia, shave, or curettage) and found that at a low setting, significant resolution of skin lesions can be achieved without significant distress to the patient. Notably, there has yet to be a recurrence of lesions two years status-post treatment with hyfrecation. Compared to previous attempted treatments, this method seems to be advantageous. It is simple, safe, cost-effective, and in our case, has shown no evidence of recurrence. In addition, there were no complications of scarring, pigment change, and/or prolonged healing that has been suggested to be associated with other treatment methods, notably laser [5]. Larger numbers of patients need to be treated prior to making any conclusions regarding this treatment method. However, these results seem promising and suggest that hyfrecation alone should be considered as a treatment alternative for the disfiguring papules associated with BHD.

## References

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