Nevus Comedonicus Syndrome

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BACKGROUND

Nevus comedonicus (NC) is a rare benign hamartomatous proliferation of pilosebaceous tissue and is clinically distinct, appearing as a patch of black keratin plugs arranged in a honeycomb pattern [1]. Therefore, diagnosis is clinical. On histology, affected hair follicles are dilated with accumulated keratin in place of the hair shaft[1,5]. There is a pyogenic variant of NC, which can result in complications such as infections, cysts, and subsequent scarring [3,5]. About 50% of NC cases are congenital, while the other 50% of cases tend to appear between the ages of 10-15 years old[5]. NC lesions primarily have cosmetic and psychosocial implications for patients.

While NC lesions are striking, the syndrome associated with NC can be less obvious. Nevus comedonicus syndrome (NCS) is a type of epidermal nevus syndrome which is classically associated with extracutaneous manifestations affecting the ocular structures, central nervous system, and skeletal system. NCS requires the presence of a NC with additional cutaneous and extracutaneous manifestations. However, there is no criteria to establish a diagnosis of NCS. Cutaneous manifestations include but are not limited to: pilar cysts and linear basal cell nevi. The most common extracutaneous manifestations have been reported to be the absence of a fifth finger and congenital cataracts (typically ipsilateral to the NS lesion). More concerning malformations tend to involve the central nervous system, with reported cases of arachnoid cysts and vascular malformations [4,6].

Nevus comedonicus and nevus comedonicus syndrome represent mosaic disorders with somatic gene mutations occurring during embryogenesis. Mutations of NEK9 and FGFR2 – which are associated with hair follicle homeostasis – remain common genetic mutations associated with NC and NCS[2].

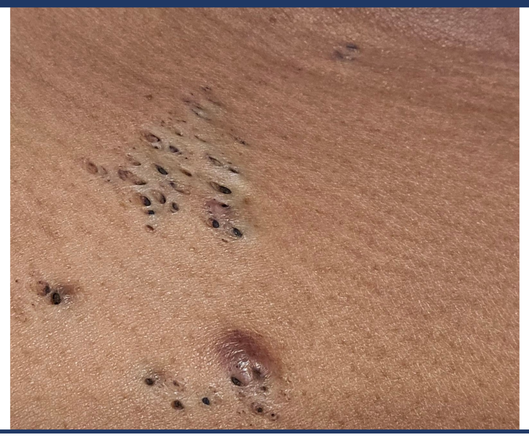
OBJECTIVE

To highlight the importance of identifying nevus comedonicus syndrome to ensure well-rounded care of these unique patients.

CASE REPORT

A 44 year-old female with a history of recurrent headaches and left congenital cataract presented for evaluation of a subcutaneous scalp nodule. During the physical exam, a 3x3cm patch of black keratin plugs arranged in a honeycomb pattern with a firm hyperpigmented 5mm subcutaneous nodule to the inferior aspect on her left upper chest was incidentally noted (*Image 1*). The lesion was consistent with a nevus comedonicus (NC). The patient recalled that the NC had been present for as long as she could remember with occasional drainage of the keratin plugs. The subcutaneous nodule on the NC had been intermittently painful and inflamed. Given the presence of NC and congenital cataract, nevus comedonicus syndrome (NCS) was suspected. A neurology referral was placed to rule out intracranial pathology, with a brain MRI pending. Single gene analysis for NEK9 was ordered, though insurance denied coverage. The patient agreed to proceed with treatment of NC and started nightly application of tazarotene. She is responding well, with plans for punch excision of the larger comedonal openings and symptomatic subcutaneous chest nodule.

Image 1. Untreated nevus comedonicus on chest of a 44-year-old female.



DISCUSSION/CONCLUSION

Even though this patient did not present to clinic for evaluation of the nevus comedonicus (NC), this incidental finding prompted further evaluation of medical history that may have initially been overlooked. Nevus comedonicus syndrome (NCS) was suspected after identification of a NC with ipsilateral congenital cataract which is one of the most common extracutaneous findings of the syndrome. With these two findings, we referred to our neurology colleagues to determine if further evaluation of the recurrent headaches was medically indicated. While genetic testing is available, its utility is unclear and should not halt medical evaluation and management of extracutaneous manifestations[2].

As there currently is no set criteria to establish diagnosis of NCS, it is important to maintain clinical suspicion in the setting of NC and be aware of the extracutaneous findings. This may be especially true in the pediatric population, as knowledge of NCS may guide practitioners toward appropriately tailored multidisciplinary care. Screening should be guided by clinical suspicion relative to medical history and symptoms to avoid unnecessary testing.

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