Could This Patch of Open Comedones Prompt Further Evaluation?

PEER REVIEWED

Photo Quiz

Volume 65- Issue 2 - February 2025

Introduction. A 44-year-old woman with a past medical history of left congenital cataract and migraines presented to a dermatology clinic with an intermittently draining lesion on the left upper chest that had been present for as long as she could remember.

History. The patient initially presented to the dermatology clinic for evaluation of skin changes on the left upper chest. Upon physical examination, there was a 3x3 cm patch of black keratin plugs arranged in a honeycomb pattern with a firm hyperpigmented 5 mm subcutaneous nodule to the inferior aspect. **(Figure 1)** Upon further questioning, she reported that the chest lesion had been present for “as long as she could remember” with occasional drainage of the black plugs and self-limited intermittent tenderness and swelling of the subcutaneous nodule.

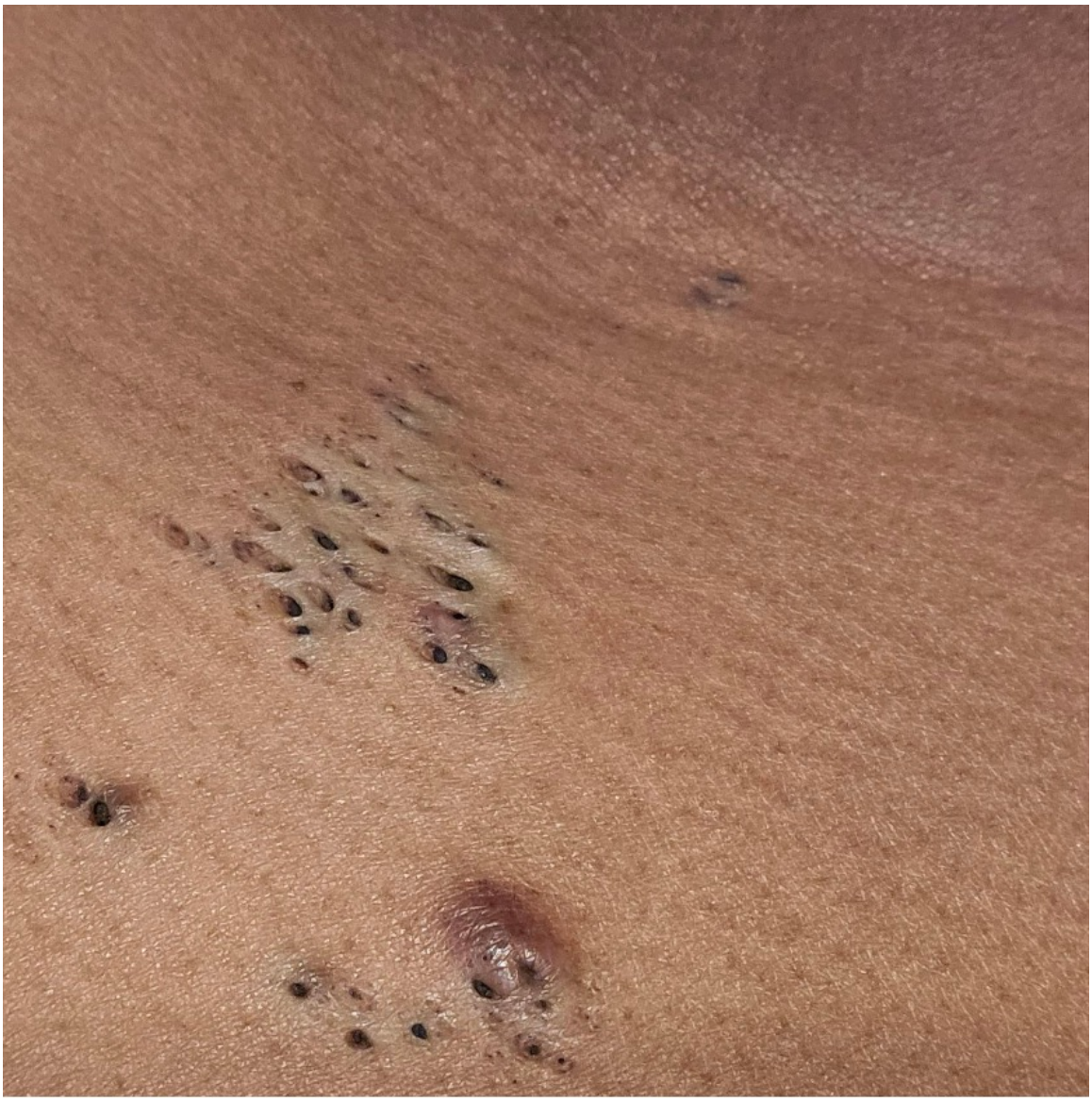

Fig. 1. *Untreated nevus comedonicus on the chest of a 44-year-old woman.*

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Correct Answer. B. Nevus comedonicus syndrome

A patch of open comedones arranged in a honeycomb pattern is consistent with a nevus comedonicus (NC). While NC lesions are striking, the syndrome associated with NC can be less obvious. In this case, the presence of an NC and history of ipsilateral congenital cataract raises concern for nevus comedonicus syndrome (NCS).

The additional answer choices listed above do not fit with the patient's history and clinical presentation. Occupational acne can present with open comedones, but usually arises at sites of exposure to cutting oils and coal tar derivatives.[6] The patient's clinical history did not fit with an exposure-related etiology given presence of comedones since childhood. Gardner syndrome is associated with multiple epidermal inclusion cysts and familial adenomatosis polyposis with an autosomal inheritance of a mutation in the APC gene.[7] Unlike NCS, the common associated ocular abnormality in Gardner syndrome is congenital hypertrophy of the retinal epithelium (CHRPE), and other associations include desmoid tumors, osteomas, dental anomalies, and thyroid tumors.8 Favre-Racouchot syndrome can have similar groupings of open comedones as NS but occurs in the setting of significant actinic damage most commonly on the face.6 Hidradenitis suppurativa (HS) usually affects intertriginous sites, and double headed comedones are pathognomonic for this condition.[7] These comedones may be accompanied by abscesses, ulcerations, sinus tracts, inflammatory nodules, and scars depending on the stage. Patients with HS may have higher rates of acne, obesity, inflammatory joint disease, inflammatory bowel disease, anxiety and depression. Patients with Down syndrome also have a predilection for developing hidradenitis suppurativa.[7] Though HS lesions can complicate NS lesions, the patient in this case has a history more suggestive of NCS.

Treatment and management. The patient was prescribed nightly application of tazarotene 0.1% cream for the NC. Given she had no further nodular swelling or pain after evaluation, she discontinued this therapy. Due to the concern for NCS in a patient with a history of recurrent migraines and ipsilateral cataract, a neurology specialist referral was placed, and the patient underwent comprehensive neurological assessment and a brain MRI without neurological abnormalities appreciated.

Outcome and follow-up. There were initial plans for topical retinoid therapy to the patient's NC and punch excision of the larger comedonal openings and symptomatic subcutaneous chest nodule. Fortunately, she had an unremarkable neurologic examination and brain MRI, which did not require further neurological evaluation. Ultimately, she chose to observe her NC instead of treatment as it remained minimally symptomatic.

Discussion. Nevus comedonicus is a rare benign epidermal nevus resulting from hamartomatous proliferation of pilosebaceous tissue.[1,2] Histologically, affected hair follicles are dilated with accumulated keratin in place of the hair shaft.[1,2,3] Clinically, NC appears as a patch of open comedones closely arranged in a honeycomb pattern.[2,3] Since the lesions are quite distinct, diagnosis is often clinically made. There is a pyogenic variant of NC, which can result in complications such as infections, cysts, and subsequent scarring.[3,4] There are even multiple reported cases of NC being complicated by hidradenitis suppurativa-like lesions.[4,5] The patient presented in this case has a pyogenic variant of NC due to the associated inflammatory subcutaneous nodule at the inferior aspect of the lesion.

Nevus comedonicus lesions primarily have cosmetic and psychosocial implications for patients. Medical management is indicated in cases involving complications from pyogenic type NC.[2] However, in cases of non-pyogenic type NC, treatment is typically cosmetic. The keratin plugs of NC can be treated with topical or oral retinoids, topical keratolytics, manual extraction, micro needling, dermabrasion, and lasers such as the neodymium YAG (Nd:YAG), CO_2, and diode.[3] Even with multiple treatment options, surgical excision seems to be the only definitive treatment with possibility of recurrence in the setting of incomplete excision.[1,2]

Nevus comedonicus syndrome is a type of epidermal nevus syndrome, which is classically associated with extracutaneous manifestations, affecting the ocular structures, central nervous system, and skeletal system.[3] NCS requires the presence of an NC with additional cutaneous and extracutaneous manifestations.[2,3] However, there is no standardized criteria to establish a diagnosis of NCS. Case reports describe a wide array of clinical presentations for NCS. Cutaneous manifestations include but are not limited to: pilar sheath tumors and basal cell carcinomas.[2] The most common extracutaneous manifestations are the absence of a fifth finger and congenital cataracts, typically ipsilateral to the NC lesion.[2,3]More concerning malformations tend to involve the central nervous system, with reported cases of arachnoid cysts and vascular malformations.[10,11] Our patient had an NC with a history of ipsilateral congenital cataract, therefore, NCS was suspected. This prompted a thorough review of systems, which elicited a history of recurrent migraines. Thus, a neurology referral was placed to rule out intracranial pathology.

About 50% of NC cases arise shortly after birth, with other cases tending to appear before the age of 10 years.[3,11] Most reported cases are likely sporadic, however, there are multiple cases of families with the lesion and one case report of homozygous twins.[12] NC and NCS represent mosaic disorders with somatic gene mutations occurring during embryogenesis.[13] Mutations of NIMA-related kinase 9 (NEK9) and fibroblast growth factor receptor 2 (FGFR2)—which are associated with hair follicle

homeostasis—remain common genetic mutations associated with NC and NCS.[2,13] While genetic testing is available, its utility is unclear and should not halt medical evaluation and management of extracutaneous manifestations.[13]

Conclusion: Although the patient's skin manifestations seemed focal and benign, it was the further investigation into her medical history that provided the information needed to suspect a genetic syndrome and ensure proper evaluation and management. 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 early in life.[10] Screening should be guided by clinical suspicion relative to medical history and symptoms to avoid unnecessary testing.

AUTHORS:
Lourdes M. López, MD[1] • Sarah Woodside, MD[2] • Jared Roberts, MD[2]

AFFILIATIONS:
[1]Department of Health Education and Training, Tripler Army Medical Center (TAMC), Honolulu, HI
[2]Department of Dermatology, San Antonio Uniformed Services Health Education Consortium (SAUSHEC), San Antonio, TX

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CORRESPONDENCE:

Lourdes M. López, MD, 1 Jarrett White Rd, TRIPLER AMC, HI 96859 (lml158@gmail.com)

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