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Case Report

Naevus comedonicus syndrome with bilateral congenital cataract and arachnoid cyst

黑頭粉刺痣綜合徵伴有雙側先天性白內障及蛛網膜囊腫

M Ozturk, H Uce Ozkol , G Dere, I An, F Caliskan Senkoy

Naevus comedonicus syndrome (NCS) is a rare syndrome with extracutaneous findings. Dilated follicular ostiums plugged with keratinous material are seen in naevus comedonicus. NCS may occur with ocular, skeletal, and central nervous system symptoms. We present this rare case of NCS which presented with bilateral blaschkoid cutaneous involvement, bilateral congenital cataract and arachnoid cyst.

黑頭粉刺痣綜合徵是一種有著皮膚以外病徵的罕見綜合徵。在黑頭粉刺痣中可見擴張的卵泡口中充塞了角蛋白物質。黑頭粉刺痣綜合徵可能出現眼部、骨骼和中樞神經系統病徵。我們介紹的這一例罕見的黑頭粉刺痣綜合徵，表現為雙側布拉許口氏樣皮膚累及、雙側先天性白內障及蛛網膜囊腫。

Keywords: Blaschkoid, cataract, naevus comedonicus syndrome

關鍵詞：布拉許口氏樣、白內障、黑頭粉刺痣綜合徵

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Introduction

Naevus comedonicus (NC) is a benign hamartoma characterised by the occurrence of comedone-like openings covered with black or brown keratin plugs, typically localised on the face, neck, arms, chest or abdomen.[1] It was first described by Kofmann in 1895.[2] NC can occur congenitally or can develop during the first decade of life. NC is a part of naevus comedonicus syndrome.[3] NC, usually has a unilateral, grouped or linear configuration. Bilateral involvement is rare.[4] We present a rare case of naevus comedonicus syndrome (NCS) with bilateral blaschkoid involvement, bilateral congenital cataract and arachnoid cyst.

Case report

A 17-year-old male patient presented to our outpatient clinic with pigmentation on the face since birth. Skin examination revealed comedone-like pores on the right side of the chest and face, on the left side of the chest and back. The lesions were located on the Blaschko lines (Figure 1). He received surgery for congenital cataract previously. Radiographic images showed no bone defect. Brain MRI showed an 11-mm arachnoid cyst near the distal part of the quadrigeminal cistern in the left deep temporal region (Figure 2). A biopsy taken from the lesion was consistent with naevus comedonicus (Figure 3). The patient was diagnosed with NCS based on dermatological, ocular and neural signs.

Discussion

NC is a disease of the epidermal naevus syndrome spectrum. It is a very rare dermatological disorder. Almost half of the cases present at birth, the remainder present during the first decade of life. Late onset is typically associated with trauma or irritation.[1] It is estimated that the prevalence of NC is between 1 in 45,000 to 1 in 100,000, without sex or race dominance. NC is usually asymptomatic and the most affected area is the face and neck, and rarely other anatomical regions, including palms, soles and genitalia, may be affected.[3] Clinically, it appears as dilated follicular ostia

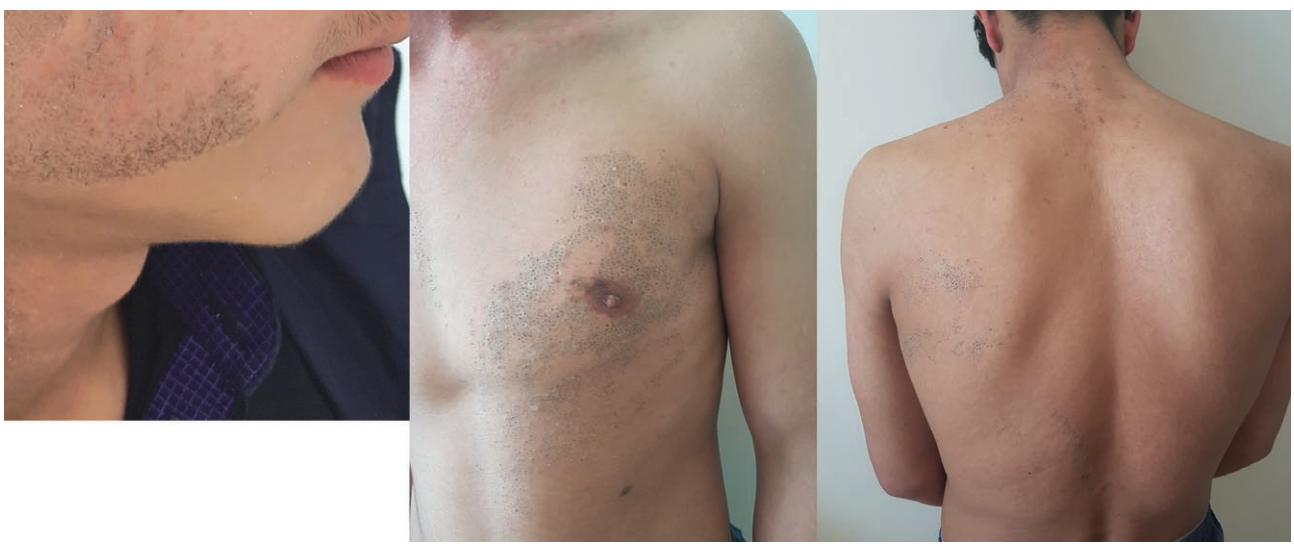

Figure 1. Blaschkoid involvement of naevus comedonicus extending to the right side of the face, neck, left chest, and upper back.

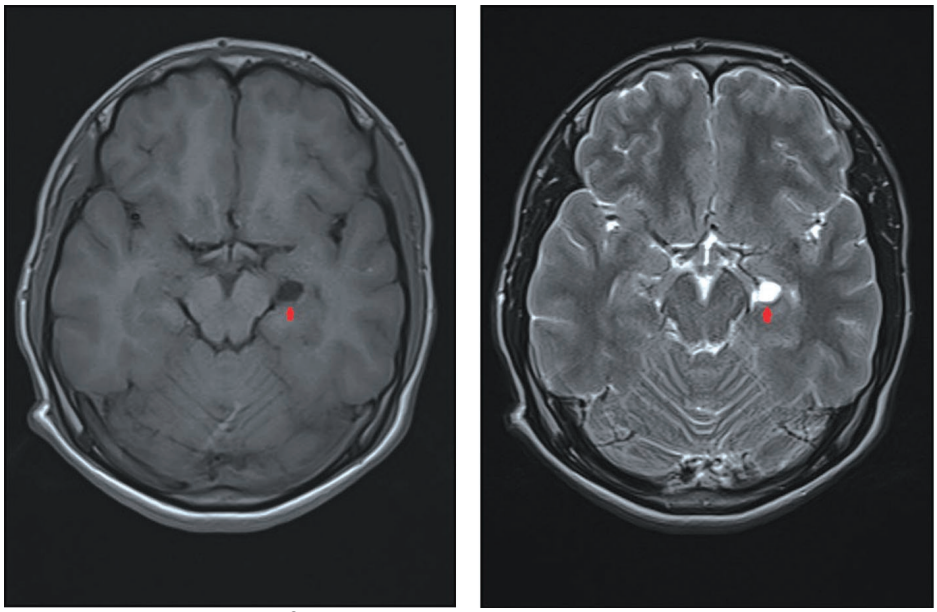

Figure 2. MRI image of arachnoid cyst (axial section T2 weight-axial section T1 weight)

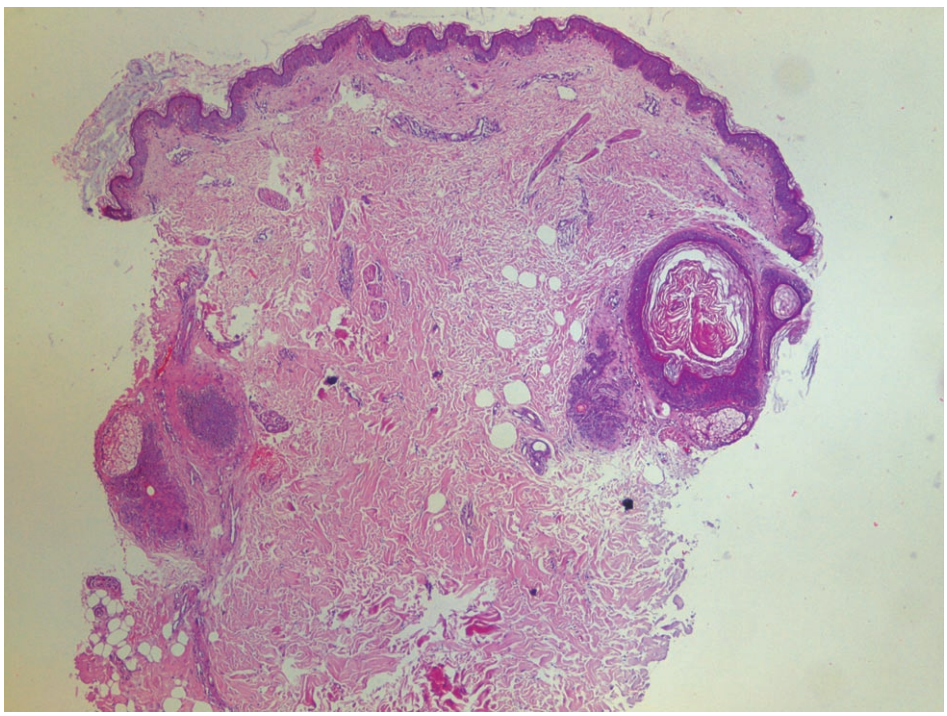

Figure 3. Skin biopsy showing invaginations of epidermis which are keratin filled and enlarged (H and E stain, x40).

plugged with pigmented keratinaceous material and may be in a linear, nevoid, bilateral or zosteriform configuration. In our case NC had blaschkoid configuration.[5]

Two types of NC are defined; the first type is non-pyogenic NC with acne-like features and the second type is characterised by the formation of papules, pustules, cysts and abscesses.[3] In NCS, extracutaneous abnormalities like Alagille syndrome, neural system abnormalities (microcephaly, Sturge-Weber syndrome, cerebral dysgenesis), multiple basal cell carcinoma, skeletal malformation (rudimentary finger, absence of fifth finger, scoliosis, trichilemmal cysts), eccrine spiradenoma and hydradenoma, bilateral follicular basal cell naevi, ipsilateral polyps and bilateral oligodontia, or ipsilateral cataract can be seen.[6] In our case, bilateral skin involvement and bilateral congenital cataract and arachnoid cyst were present.

NCS is a disease of epidermal naevus syndrome including angora hair naevus syndrome, Becker naevus syndrome, phakomatosis pigmentokeratotica and Schimmelpenning syndrome.[3] The exact genetic basis of NC is unknown. In a linear NC case, a fibroblast growth factor receptor 2 (FGFR2) mutation was found. Germ cell mutations in FGFR2 lead to Apert syndrome characterised by cystic acne and severe comedonal lesions.[7] Characteristic histopathological findings of NC are enlarged follicles surrounded by atrophic epithelium and filled with orthokeratotic horny substance. Interfollicular epidermis is usually papillomatous and hyperkeratotic with small sebaceous glands and scattered hair follicles seen in the early stage of NC lesions.[8] Differential diagnosis includes other linear eruptions such as epidermal naevus, lichen striatus, porokeratosis, lichen planus, lipoid proteinosis or tattoo reactions.[4]

Aggressive therapies are not necessary as NC is benign. Treatment can be given for cosmetic reasons. Topical retinoic acid and 12% ammonium lactate may be used. Tacalcitol and tazarotene have been effective in combination with calcipotriene 0.05% and 0.005% applied daily although recurrences are frequent. Oral retinoids like isotretinoin have been ineffective in most cases, but they can be an option if there is systemic involvement. Due to their antivascular activity, FGFR inhibitors may be a treatment option.[3,5] In our patient, the keratin plugs resolved with tretinoin cream.

To the best of our knowledge, this is the first case of NCS with arachnoid cyst and bilateral congenital cataract in the English literature. In cases of NC, the possibility of extracutaneous involvement should be kept in mind.

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Conflicts of interest

There are no conflicts of interest.

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