OLGU SUNUMU CASE REPORT

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Nevoid Basal Cell Carcinoma Syndrome (Gorlin-Goltz Syndrome): A Patient Showing Distinctive Dermoscopic Features: Case Report

Nevoid Bazal Hücreli Kanser Sendromu (Gorlin-Goltz Sendromu): Özgün Dermoskopik Bulguları Yansıtan Bir Olgu

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ABSTRACT Nevoid basal cell carcinoma syndrome, which is also known as Gorlin-Goltz syndrome, is an autosomal dominant disorder characterized by a wide range of clinical manifestations, including multiple early-onset basal cell carcinomas, palmoplantar pits and skeletal deformities. Although nevoid basal cell carcinoma syndrome is a rare genodermatosis, since its first description in 1960 by Gorlin and Goltz, there have been several case reports delineating diverse clinical manifestations of this unique disease. On the other hand, there have been fewer reports on the dermoscopic features of basal cell carcinomas found in these patients. Here in this case report, we present a case with nevoid basal cell carcinoma syndrome, who demonstrates typical clinical features also displays striking dermoscopic findings of basal cell carcinomas, which are the most significant manifestation of the syndrome.

Keywords: Basal cell nevus syndrome; basal cell carcinoma, multiple; dermoscopy

ÖZET Gorlin-Goltz sendromu olarak da bilinen nevoid bazal hücreli karsinom sendromu, erken yaşta gelişen çok sayıda bazal hücreli kanser, palmoplantar çukurcuklar ve iskelet sistemine ait deformiteleri de içeren oldukça geniş klinik manifestasyonlarla karakterize otozomal dominant bir hastalıktır. Nevoid bazal hücreli karsinom sendromu her ne kadar nadir bir genodermatoz olsa da, ilk defa 1960 yılında Gorlin ve Goltz tarafından tanımlanmasından bu yana, hastalığın farklılık gösteren klinik bulgularını yansıtan birçok olgu bildirisi şimdiye kadar yayımlanmıştır. Bununla birlikte, bu hastalardaki bazal hücreli kanserlerin dermoskopisini yansıtan yayınlara daha az rastlanmaktadır. Biz de bu olgu sunumu ile, nevoid bazal hücreli karsinom sendromunun tipik klinik bulgularını yansıtan, aynı zamanda hastalığın en önemli manifestasyonu olan bazal hücreli kanserlerin dikkat çekici dermoskopik bulgularını da ortaya koyabildiğimiz bir olguyu sunmak istiyoruz.

Anahtar Kelimeler: Bazal hücreli nevüs sendromu; bazal hücreli karsinoma, multipl; dermoskopi

Nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin-Goltz syndrome, is a rare genetic disorder with multiple and varied clinical manifestations. Although NBCCS was first recognized by Jarisch and White in 1894, it was not until 1960s that Gorlin and Goltz more clearly defined the syndrome as "multiple naevoid basal cell epithelioma, jaw cysts and bifid rib syndrome."[1-3] Since then, several reports have been published in the literature that demonstrate and evaluate clinical manifestations of NBCCS.[1-12] However, there have been fewer reports describing dermoscopic features of basal cell carcinomas found in these patients.[13-20] Here, we present a case of NBCCS, which also presents dermo-

scopic features of basal cell carcinomas found in this syndrome.

CASE REPORT

A 30-year-old male patient came to our outpatient clinic with a several years history of multiple enlarging lesions on his face and trunk. While his past medical history was unremarkable, his family history revealed the evidence of similar lesions in his grandfather, who had died several years ago. Upon examination, we observed that the patient had frontal bossing and kyphoscoliosis (Figure 1). Dermatological examination revealed multiple dark coloured, irregularly bordered papules ranging in size from 3 to 10 mm over his face (Figures 1-3). On his left middle back a brownish asymmetrical plaque and on the middle abdominal region a heavily pigmented nodule (Figure 4) were detected. We also noticed few palmar however numerous plantar pits (Figure 5). On dermoscopic examination typical features of basal cell carcinoma, which are absence of pigment network, maple-leaf like structures, arborizing vessels, blue-grey ovoid nests, blue-grey globules and dots, concentric structures, spoke-wheel structures and ulceration (Figures 6-16) were identified [MoleMax I Plus® (Derma Medical Systems GmbH, Vienna, Austria)]. Histopathological examination of the lesions from the abdominal area, back and his neck was consistent with basal cell carcinoma. Computed tomography images revealed calcification of bilateral basal ganglia, falx cerebri and tentorium cerebelli, also odontogenic keratocysts. The patient was referred to neurology and plastic surgery department and the treatment of the patient was planned as oral vismodegib 150 mg daily.

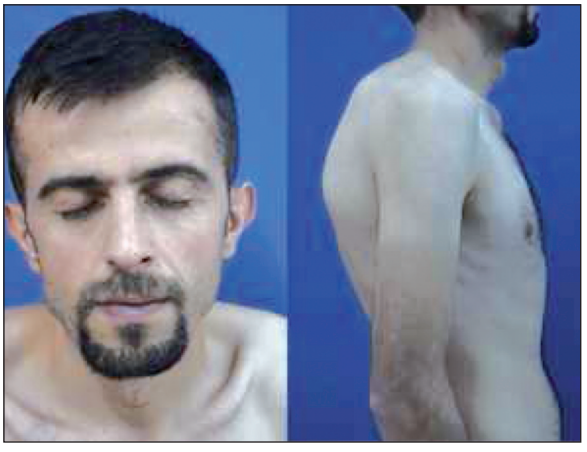

FIGURE 1: Frontal bossing, multiple dark coloured lesions over the face and kyphoscoliosis.

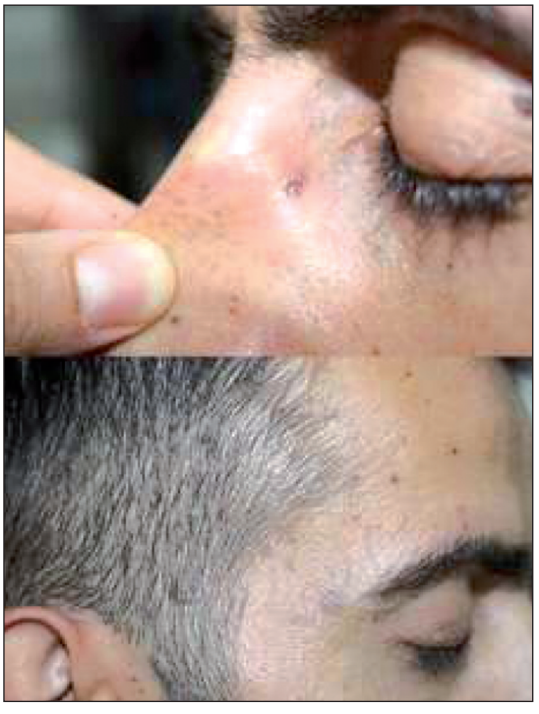

FIGURE 2: Peripheral foci of blue-grey pigment on a translucent background on the dorsum of the nose (upper image), multiple dark coloured lesions over the face and scalp (lower image).

DISCUSSION

Nevoid basal cell carcinoma syndrome (NBCCS) is a rare genodermatosis, characterized by a wide range of clinical manifestations. NBCCS is also implicated with increased mortality and morbidity because of its association with neoplasms. It is an autosomal dominantly transmitted disorder with variable expressivity, that is the reason why all clinical findings do not appear in all patients. NBCCS is a multisystemic disease, however the most striking finding of the syndrome is the copious number of basal cell carcinomas, which is the origin of the term nevoid basal cell carcinoma syndrome.[1-12,21-23]

Basal cell carcinomas emerge in various stages of the syndrome. In the literature although there

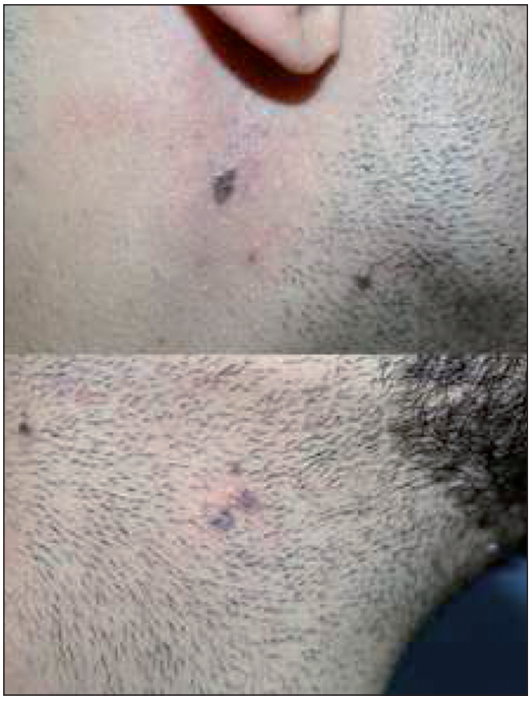

FIGURE 3: Dark coloured greyish millimetric papules coalescing to form an oval shaped lesion and a dome shaped papule on the inferolateral aspect (upper image), greyish irregularly bordered lesions with a translucent appearance on the right side of the neck (lower image).

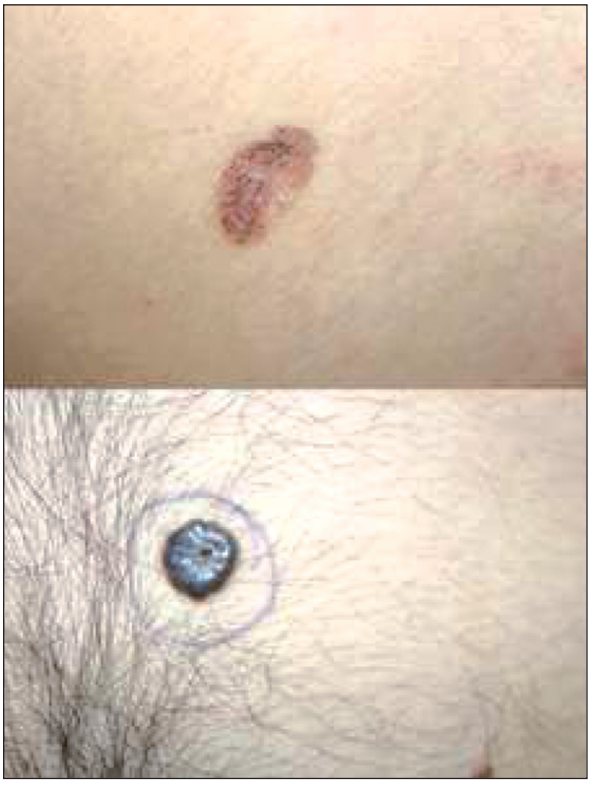

FIGURE 4: An asymmetrical irregularly bordered brownish plaque intermingled with darker millimetric papules (upper image), a heavily pigmented nodule with a central depression (lower image).

are cases in pediatric ages, most commonly basal cell carcinomas appear between puberty and 35 years of age. The median age of onset for the appearance of basal cell carcinomas has been reported to be 25.[3,7] Cancers also vary in number from few to multitute and there is an obvious relationship between the number of cancers and sun exposure. Most common initial site is the nape of the neck and sun-exposed areas including, face, the back and the chest are the typical locations. Usually they range in size from 1 to 10 mm in diameters. Characteristics of basal cell carcinomas of this syndrome do not differ from characteristics of sporadic ones. They manifest as pearly, flesh-coloured papules, plaques or noduloulcerative lesions. They rarely metastasize and most of them do not show progression. It has been reported that it is only after puberty that the basal cell carcinomas can become aggressive.[1-12,21-23]

The diagnosis of NBCCS relies on a set of defined clinical criteria. Major diagnostic criteria include multiple (>2) basal cell carcinomas or one under 20 years, odontogenic keratocysts of the jaws

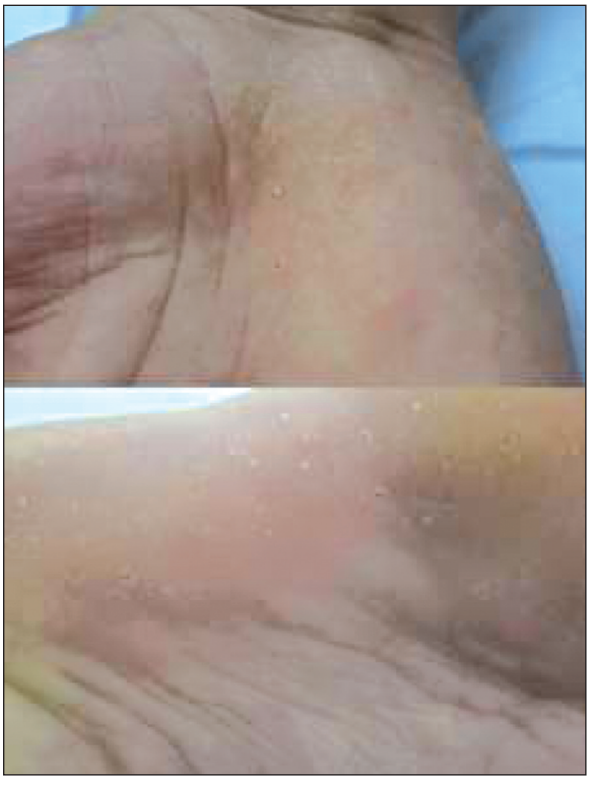

FIGURE 5: Palmoplantar pits.

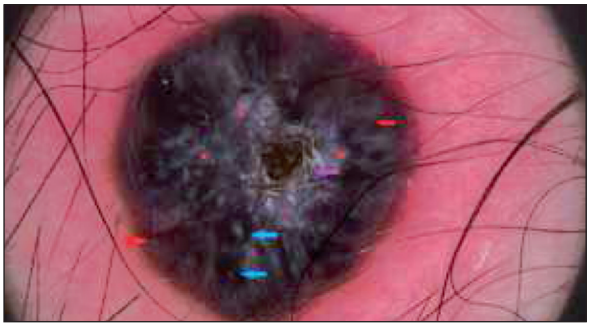

FIGURE 6: Dermoscopic image of the lesion in Figure 4; multiple large blue grey ovoid nests (blue arrows), an ulceration with a bloody crust (violet arrow), arborizing vessels (red arrows), white structureless areas showing dermal fibrosis (red stars).

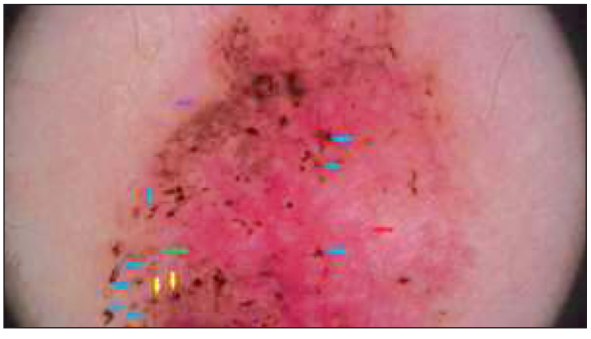

FIGURE 7: Dermoscopic image of the lesion in Figure 4 (upper segment); absence of pigment network, fine telangiectasias (red arrow), leaf-like areas (green arrow), blue-grey dots (violet arrow), concentric structures (blue arrows), spoke-wheel areas (yellow arrows).

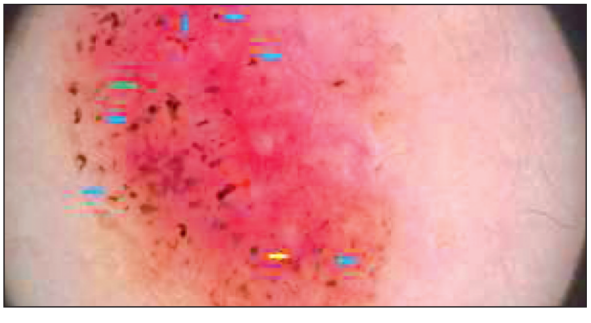

FIGURE 8: Dermoscopic image of the lesion in Figure 4 (lower segment); leaf-like areas (green arrow), concentric structures [blue arrows (precursors of spoke-wheel areas)], ulceration (red star), fine telangiectasias (prominent on the middle right border of the lesion), blue-grey dots (yellow arrow)

proven by histopathology, palmar or plantar pits (3 or more), bilamellar calcification of the falx cerebri, bifid, fused or markedly splayed ribs and first degree relatives with NBCCS. Minor diagnostic criteria include macrocephaly, congenital malformation, skeletal abnormalities, radiological abnormalities, ovarian fibroma and medulloblastoma. In the presence of two major criteria or one major and two minor criteria, the diagnosis of NBCCS is made.[7] Our patient had frontal bossing, kyphoscoliosis, multiple basal cell carcinomas and palmoplantar pits, also computed tomography images revealed calcification of bilateral basal ganglia, falx cerebri and tentorium cerebelli, also odontogenic keratocysts. Therefore, he fulfils the diagnostic criteria for NBCCS. Since our patient does not show other features of NBCCS, we do not discuss the remaining clinical manifestations of the syndrome. On the other hand, we planned to follow up the patient for clinical evidence of any tumor, including medulloblastoma and others.

Dermoscopy is a non-invasive, practical imaging method that allows in vivo evaluation of pigmented and nonpigmented skin lesions.[24] Dermoscopy appears to be a quick and efficient tool in the early detection of basal cell carcinomas in NBCCS. In the literature, there have been reports

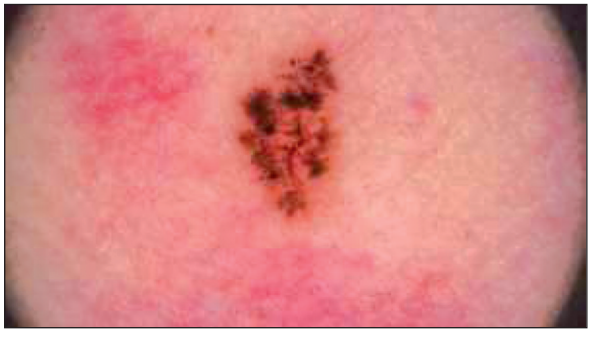

FIGURE 9: Spoke-wheel areas

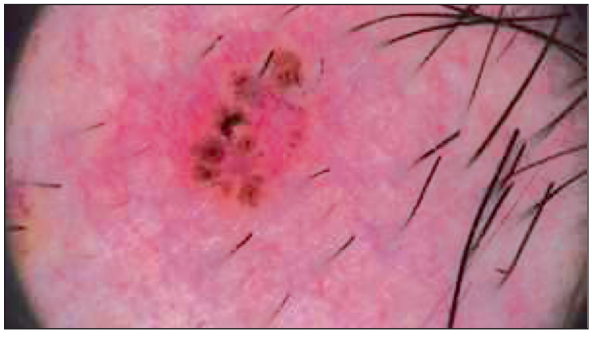

FIGURE 10: Concentric structures with darker central areas.

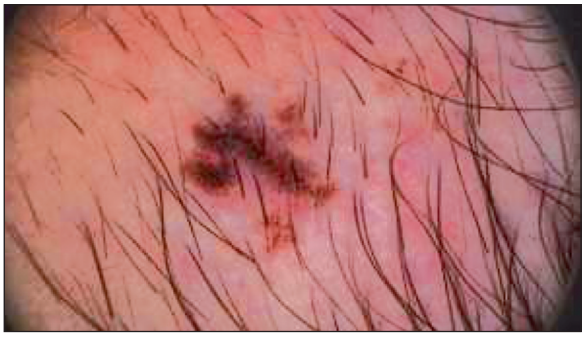

FIGURE 11: Multiple blue-grey globules and dots

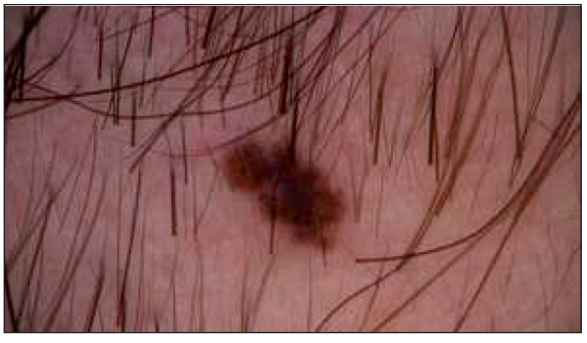

FIGURE 12: Absence of pigment network, multiple blue-grey dots and globules.

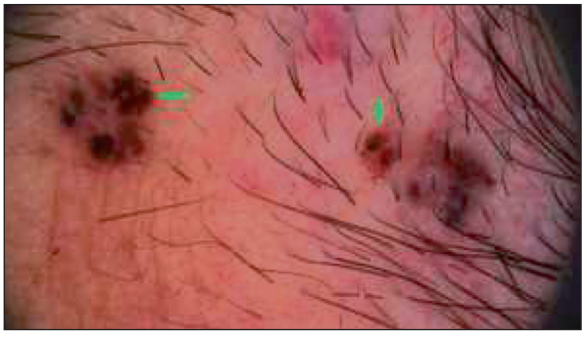

FIGURE 13: Leaf-like areas.

describing dermoscopic features of basal cell carcinomas found in NBCCS.[13-20] It is known that typical dermoscopic features of sporadic basal cell carcinomas include blue grey ovoid nests, blue grey globules, spoke-wheel areas, arborizing telangiectasias, maple-leaf like areas and ulceration. Since characteristics of basal cell carcinomas of NBCCS do not differ from characteristics of sporadic ones, it is not unpredictable to detect typical dermoscopic features including spoke-wheel areas, maple-leaf like areas, blue grey ovoid nests and ulceration in basal cell carcinomas of NBCCS.[13-20] Moreover, it has been suggested that maple leaf-like areas, short fine superficial telangiectasia, multiple small erosions, and shiny white-red structureless areas seem to be potent predictors of superficial basal cell carcinomas, whereas the presence of arborizing vessels, blue grey ovoid nests and ulceration seem to be related with non-superficial basal cell carcinomas of NBCCS.[19] We have also confirmed these findings and demonstrated that dermoscopic features of the pigmented nodule

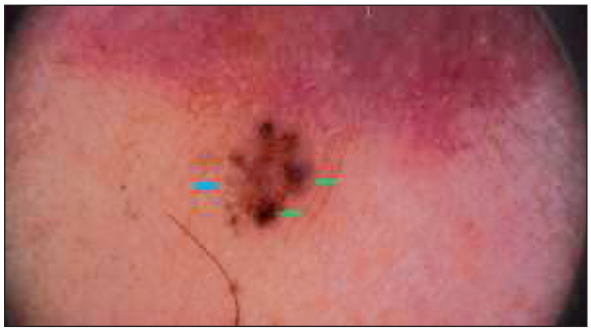

Figure 14: : Leaf-like areas (green arrows), multiple blue-grey dots (blue arrow).

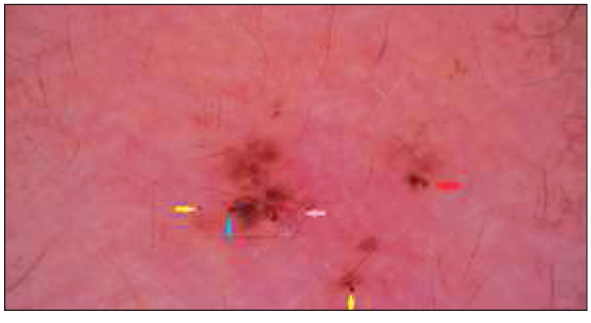

Figure 15: Concentric structure (red arrow), arborizing vessel (pink arrow), blue-grey nests (blue arrow), blue-grey dots (yellow arrows)

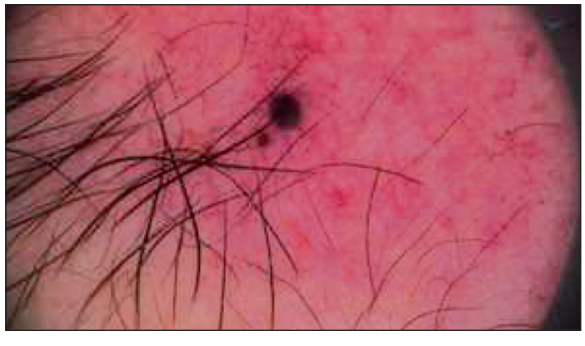

Figure 16: Large blue-grey ovoid nests.

on the abdominal region of the patient included multiple large blue grey ovoid nests, arborizing vessels and an ulceration. On the other hand, dermoscopic findings of the superficial basal cell carcinoma found on the back of the patient composed of mainly fine telangiectasias, leaf-like areas and spoke-wheel areas. Dermoscopy is helpful not only in the early detection of basal cell carcinomas of NBCCS, but also in the management of the affected patients.[13-20] We suggest that further reports and studies are needed on the dermoscopic findings of basal cell carcinomas of NBCCS.

In this case report we present a case with NBCCS. NBCCS is a multisystemic disease with malignant potential. Our patient manifests skeletal deformities, radiological findings and cutaneous features of the syndrome, most importantly basal cell carcinomas. Hence, we highlight the outstanding findings of our patient. On the other hand, as far as we know, there are few reports in the literature presenting dermoscopic findings of these patients. Dermoscopic findings of our patient were striking and we assume that further case reports describing similar features are needed.

Conflict of Interest

Authors declared no conflict of interest or financial support.

Authorship Contributions

Concept: *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Design:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Supervision:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Funding:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Materials:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Data Collection and/or Processing:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Analysis and/or Interpretation:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Literature Review:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Writing:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın;* ***Critical Review:*** *Ahu Yorulmaz, Ahmet Uğur Atılan, Başak Yalçın.*

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