A rare presentation of Gorlin syndrome in a patient of African ancestry

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Background/Introduction

Gorlin syndrome is a rare genetic disease that is also referred to as Nevoid Basal Cell Carcinoma Syndrome (NBCCS). The syndrome is caused by an autosomal dominant mutation in the SUFU or patched (PTCH1) gene, which has variable expressivity but complete penetrance. The features of this disorder include numerous basal cell carcinomas (BCCs) over the course of the patient's life starting in youth, frontal bossing, macrocephaly, skeletal abnormalities, palmar plantar pits, postnatal tumors, medulloblastoma, and odontogenic keratocysts of the mandible, and calcification of the falx cerebri.[1-3] Given the constellation of symptoms, patients are often diagnosed early in life with this syndrome. Patients without complete expressivity of the development of these symptoms may have a delay in diagnosis. Furthermore, patients of color with this syndrome may also have a delay in care because of how rare the condition is and the unfamiliar presentation in this population.

Case Report

A 57-year-old Black female with past medical history of BRCA+ triple negative invasive ductal breast carcinoma presented with numerous hyperpigmented smooth and well-demarcated plaques on the scalp, neck, back, and lower extremities (**Figure 1)**.

She was referred to our dermatology clinic after breast cancer genetic screening. She was found to have both an intronic variant of unknown significance for PTCH1 gene and BRCA1 gene. Personal and family history were negative for skin cancer or dental issues. Patient reported a non-healing lesion on her scalp for an unknown amount of time while undergoing chemotherapy. A few years ago, she had another non-healing lesion on her scalp and was encouraged by her beautician to go to a dermatologist to have it evaluated. However, she never saw a dermatologist and attributed it to irritation from periodic hair relaxers for several years. She was also told that she had "fragile" areas in the jaw on X-ray when she enlisted in the military reserve at 17-years-old but has not had follow-up imaging.

Physical examination revealed four 1.5-3 centimeters plaques with black papules and macules on the anterior and posterior scalp (**Figure 2 & 3**). Under dermoscopy, there was a background of a pink hue with multiple Maple Leaf-patterned black pigmented globules and pink and black spicules. Shave biopsies of all four lesions were positive for basal cell carcinomas (BCCs). The plaques were histologically characterized by junctional melanocytic nevus with moderate cytologic atypia. There were two discrete palmar pits (**Figure 4**) on the one of the left fourth digit and left palm. She did not have frontal bossing or hypertelorism.

She was referred to Mohs surgery for treatment and we discussed the need for daily sunscreen use and continued close monitoring with skin exams.

Figure 1 . Numerous Basal Cell Carcinomas along the scalp

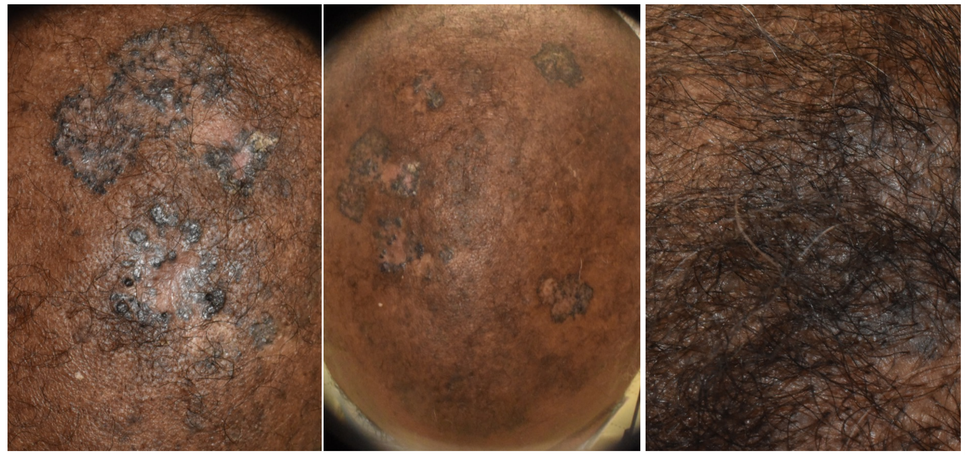


Figure 2. Close-ups of Basal Cell Carcinoma

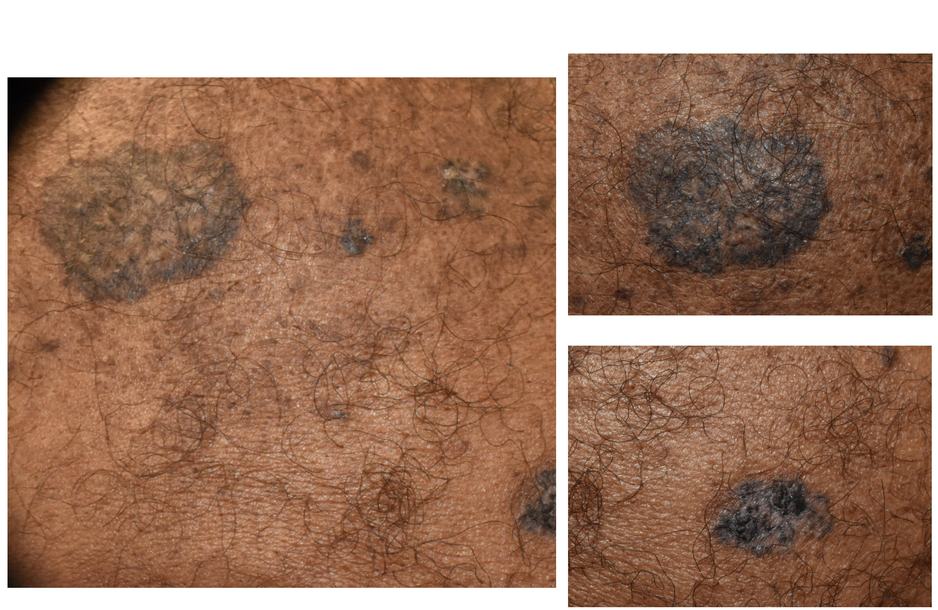


Figure 3. Scalp

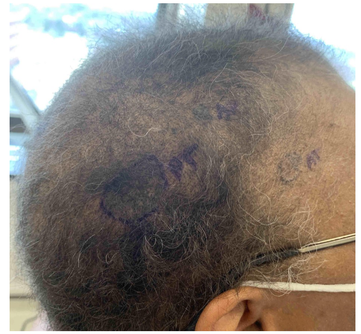
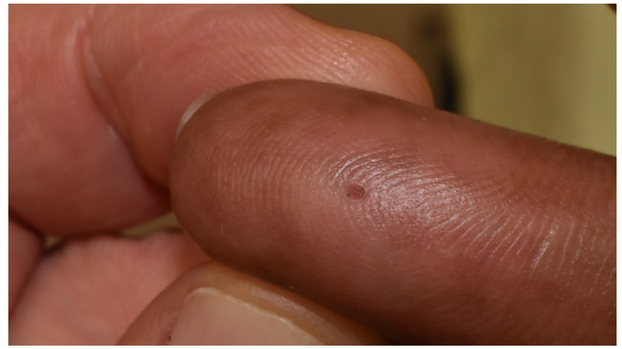


Figure 4. Palmar Plantar Pits



Discussion

This case highlights the delayed diagnosis of Gorlin syndrome in a patient of color in an effort to enable recognition of this unique constellation of systems that is not well documented in darker skin tones. Although a rare disorder, patients with this syndrome should be monitored long term for malignancy and there should be a high suspicion to biopsy persistent lesions that may clinically appear as benign. There may be a delay in diagnosis of Gorlin syndrome in patients with skin color given there are limited reports in the literature of this genodermatosis in patients with darker skin tones. These lesions may be mistaken for SKs, benign nevi, scars, etc. Providers should be aware that this syndrome occurs in patients of color.

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Disclosures: The authors have no disclosures.

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