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Hermansky-Pudlak Syndrome

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Clinical Image

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A 58 year-old man from Puerto Rico presented to our clinic in September 2018 for multiple nonhealing wounds on the face and scalp. Physical examination was remarkable for bright blue eyes, numerous lentigines, actinic damage, and decreased pigmentation of the skin, hair, eyebrows, and eyelashes (Figures 1–3). The patient stated that his hair and skin have been lightly pigmented since birth and began developing brown pigmented lesions during childhood. His past medical history was also remarkable for multiple nonmelanoma skin cancers, easy bruising, nystagmus, and a diagnosis of pulmonary fibrosis in his late 40s that required lung transplantation. Given the patients physical examination findings, he was diagnosed with Hermansky-Pudlak syndrome (HPS). Chemoprevention with nicotinamide was initiated, and he was monitored with regular skin examinations every 3 months thereafter. He was advised to avoid aspirin and NSAIDs given the high risk of platelet dysfunction associated with his condition. The patient was closely followed by our clinic and treated for multiple cutaneous squamous cell carcinomas over the following 2 years, but he was then lost to follow-up.

HPS is an autosomal recessive multisystemic disorder characterized by oculocutaneous albinism, bleeding diathesis, and the involvement of various internal organs [1]. HPS is seen with higher frequency in Puerto Rican patients, which is believed to be due to a founder effect [2]. Genetic mutations in HSP result in defective biogenesis of melanosomes and lysosome-related organelles [1]. Systemic complications may include granulomatous colitis, pulmonary fibrosis, and cardiomyopathy as a result of lysosomal ceroid accumulation. Bleeding diathesis is due to a platelet storage pool defect and may manifest as ecchymoses, nosebleeds, and menorrhagia. Platelets will classically demonstrate an absence of dense bodies on

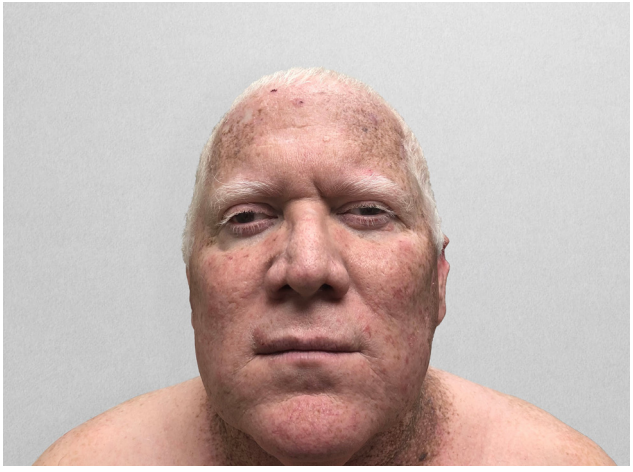

Figure 1: Blue eyes and decreased pigmentation of the skin, hair, eyebrows, and eyelashes. There are numerous coalescing brown macules and erythematous scaly papules in sun-exposed regions of the face and chest.

Figure 2: Decreased pigmentation of the skin, hair, eyebrows, and eyelashes. There are numerous coalescing brown macules and erythematous scaly papules in sun-exposed regions of the face.

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Figure 3: Blue eyes and decreased pigmentation of the eyebrows and eyelashes.

electron microscopy. Ocular findings may include pale irides and early nystagmus. Cutaneous malignancies are common. The most common cause of death is pulmonary fibrosis, with a life expectancy of 30–50 years [3]. Frequent skin examinations, annual pulmonary function tests beginning at age 20, and avoidance of aspirin or NSAIDs are an essential aspect of the long-term management [3].

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Informed consent: The patient described in this report provided written informed consent.

References

1. Huizing M, Helip-Wooley A, Westbroek W, Gunay-Aygun M, Gahl WA. Disorders of lysosome-related organelle biogenesis: clinical and molecular genetics. Annu Rev Genom Hum Genet 2008; 9:359–86.
2. Toro J, Turner M, Gahl WA. Dermatologic manifestations of Hermansky-Pudlak syndrome in patients with and without a 16-base pair duplication in the HPS1 gene. Arch Dermatol 1999;135: 774–80.
3. El-Chemaly S, Young LR. Hermansky-Pudlak syndrome. Clin Chest Med 2016;37:505–11.