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This landmark article by Frantisek Hermansky and Paulus Pudlak, clinicians in Prague, Czechoslovakia, is the first to describe 2 unrelated individuals with what is now called Hermansky-Pudlak syndrome, a bleeding disorder that occurs in association with oculocutaneous albinism. The definition of this syndrome resulted not only in improved care of these patients but also in a functional and molecular understanding of the disease and the role of dense granule secretion in platelet function. Hermansky-Pudlak syndrome is now known to be related to defective dense granule biogenesis due to mutations in any of ≥9 different genes.

Hermansky F, Pudlak P. Albinism associated with hemorrhagic diathesis and unusual pigmented reticular cells in the bone marrow: report of two cases with histochemical studies. *Blood*. 1959; 14(2):162-169.

Albinism Associated with Hemorrhagic Diathesis and Unusual Pigmented Reticular Cells in the Bone Marrow: Report of Two Cases with Histochemical Studies

By F. Hermansky and P. Pudlak

HEMORRHAGIC DISORDERS with a prolonged bleeding time as a main laboratory finding and a normal platelet count may have a varying pathogenesis. They are usually caused by qualitative changes of the platelets or a disturbance of the vascular factor in the process of hemostasis.[29] They are for the most part congenital and hereditary anomalies, which, despite the progress made in this field, are still difficult to classify into well-defined groups.[2,20] We had the opportunity of observing a hemorrhagic disorder of medium severity with a prolonged bleeding time in two unrelated albinos. These observations were made independently in two separate institutions. Both cases had concomitant congenital nystagmus, and in both, large reticular cells with an unusual pigment were found in the bone marrow, which could not be identified with reference to similar cells described in the literature.[15,24] The combination of these abnormalities independently observed in two unrelated patients suggested that we were confronted with a new, not previously described congenital anomaly.

Case Studies

Case 1. E.B., a 33-year-old children's nurse, was first admitted to the First Medical Clinic in Prague on December 22, 1953 because of severe, recurrent epistaxis not related to menstruation. From childhood the patient had noticed that she bruised easily and tended to bleed for a long time after injury. Severe menorrhagia had occurred from time to time since puberty, simultaneously with increased bruising and more frequent epistaxis. In 1951, she had severe bleeding after a dental extraction. During her several admissions to the hospital she was given repeated blood transfusions with doubtful hemostatic effect. Otherwise, the past history was not contributory and there was no family history of hemorrhagic disease or albinism.

The clinical examination showed a generalized lack of pigment in the skin and hair. The iris was a yellowish-green, and was reddish on lateral illumination. Furthermore, there was strabismus and continuous coarse nystagmus. There were scattered ecchymoses on the skin of the extremities. Otherwise, the clinical findings were within normal limits.

The blood pressure was normal, 130 90. There were subfebrile to febrile temperatures. The chest x-ray showed no focal changes. Ophthalmologic examination revealed myopia and confirmed the presence of albinism with oculogenic nystagmus.

Laboratory findings: Urinalysis, negative; increased sedimentation rate, 51 mm. hr.; Wassermann reaction, WR/ negative. Flocculation reactions: Weltmann 7; Takata negative; thymol turbidity test, 7.8 units; cholesterol, 214 mg. per cent. Serum bilirubin: total 0.8 mg. per cent, direct negative. Blood cultures were repeatedly negative. TB-

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