

Hermansky-Pudlak / Chediak- Higashi Syndrome.

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Received date: December 05, 2023; Accepted date: January 15 2024; Published date: January 22, 2024

Citation: H.D. Solomons, (2024), Hermansky-Pudlak / Chediak- Higashi Syndrome, Clinical Oncology Case Reports, 3(1); DOI: 10.31579/ 2834-5061/14

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Abstract

The common denominator is albinism.Hermansky Pudlak syndrome affects the platelets and patients have a tendency to bleed.(1.)

Keywords: chediak-higashi disease; lysosomal; immune system

Introduction

The common denominator is albinism.Hermansky Pudlak syndrome affects the platelets and patients have a tendency to bleed.(1.)

Chediak-Higashi syndrome affects the leukocytes, results in immune disorders and has intracytoplasmic inclusions. These latter patients are prone to malignant lymphomas as the immune system is involved.

Hermansky-Pudlak symptoms occur due to defects in melanosomes and affects lysosomal organelles in cells esp., platelet dense granules.For this reason these patients have a haemorrhagic tendency and tend to bleed.

Chediak-Higashi syndrome patients usually die at an early age. It also affects lysosomal organelles. (2.)

Chediak-Higashi is an autosomal recessive disorder as is Hermansky-Pudlak syndrome. 3 subtypes of Hermansky-Pudlak disease exist.

Chromosomes 3,5 and 10 are involved.

Hermansky-Pudlak is seen predominantly in Puerto Ricans but is also found in the Swiss Alps.

In Chediak-Higashi disease 8 known gene allele defects are found, natural killer cells are deficient and the immune system is involved predisposing them to lymphomas.

In both disorders' hair, skin and eye colour are deficient making albinism the common factor! (3.)

Thus, it can be concluded that Hermansky-Pudlak/Chediak-Higashi Syndrome affects the platelets and the white cells; viz., the haematological system.

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