Hermansky-Pudlak Syndrome

Group of inherited diseases

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Hermansky-Pudlak syndrome is the name given to a group of inherited diseases that have symptoms of albinism, bleeding problems, and cellular storage disorders. Hermansky-Pudlak syndrome is estimated to occur in 1 in 1,800-2,000 individuals in Puerto Rico, where there is the highest concentration of individuals with the syndrome. Hermansky-Pudlak syndrome has also been identified in a village in the Swiss Alps and in persons of Dutch, Turkish, Pakistani, and Japanese ethnicity.

Seven gene defects are associated with the four known subtypes of Hermansky-Pudlak syndrome. Some individuals develop the syndrome but do not have any of the known gene defects, so more research needs to be done in this area.

Symptoms

There are three main disorders caused by Hermansky-Pudlak syndrome, which result in these symptoms:

Albinism and eye problems - Individuals will have varying amounts of skin pigment (melanin). Because of the albinism there are eye problems such as light sensitivity (photophobia), strabismus (crossed eyes), and nystagmus (involuntary eye movements). Hermansky-Pudlak syndrome also impairs vision.

Bleeding disorders – Individuals with the syndrome have platelet dysfunction. Since platelets are necessary for blood clotting, individuals will bruise and bleed easily.

Cellular storage disorders – Hermansky-Pudlak syndrome causes a wax-like substance (ceroid) to accumulate in the body tissues and cause damage, especially in the lungs and kidneys.

Diagnosis

Hermansky-Pudlak syndrome may be diagnosed in infancy from the presence of albinism. A bleeding disorder may be noted in a boy who is circumcised; otherwise, as the individual grows he or she may have frequent nosebleeds, bleeding gums, and frequent bruising.

Ceroid may collect in the intestines and cause symptoms of diarrhea, cramps, and bloody stools similar to inflammatory bowel disease. Ceroid in the lungs may cause shortness of breath.

If an individual is suspected of having Hermansky-Pudlak syndrome, his family history will be investigated for relatives having similar symptoms or having Puerto Rican ancestry. Genetic testing can be done for one of the defective genes associated with the syndrome. However, the definitive test used to detect Hermansky-Pudlak syndrome is the examination of platetlets under an electron microscope.

Treatment

Treatment for Hermansky-Pudlak syndrome focuses on minimizing the complications of bleeding, monitoring the lungs and kidneys for damage by ceroid, and frequent skin checks by a dermatologist to watch for skin cancer. A computed tomography (CT) scan can look for any hidden internal bleeding as well as check for organ damage. Avoiding smoking and receiving prompt treatment for any respiratory infections will promote healthy lungs. Low vision aids, physical therapy, and frequent doctor check-ups will also promote health. Unfortunately, many people with Hermansky-Pudlak syndrome die from its complications, especially lung damage.

Genetic counseling

Hermansky-Pudlak syndrome is inherited in an autosomal recessive manner, meaning that both parents must be carriers, or actually have the disease, for a child to develop the syndrome. Each child born to the parents will have a one in four chance of inheriting the syndrome.

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