Hermansky-Pudlack Syndrome

Main Features

- Lethal subtype of Oculocutaneous albinism
- Tyrosinase-positive oculocutaneous albinism
    - Skin can be from white to olive
    - Hair color can be from white to brown
- **Bleeding diathesis** from platelet storage pool deficiency
- **Pulmonary Fibrosis**
- **Granulomatous colitis**

Eye Findings

- Iris pigment abnormalities with iris transillumination
- Reduced retinal pigment
- Foveal hypoplasia
    - VA usually between 20/50 to 20/400
    - Nystagmus
    - Abnormal increase in crossing of optic nerve fibers at the optic chiasm

Diagnosis

- Clinical findings above
- Platelet electron microscopy shows absent dense bodies
    - Most often used to confirm diagnosis
- Increased bleeding time
- Impaired platelet aggregation
- Urinary ceroid lipofuscin deposits is characteristic but virtually never used for diagnosis
- Molecular diagnosis available,
    - Testing for HPS 1-8 available clinically
    - HPS 9 (BLOC1S6, PLDN) is available on a research basis only

Management

- Correct Refractive Error
- Protect skin from the sun
- Protracted bleeding with surgery- appropriate treatment needed
    - gelfoam, DDAVP, platelet transfusions
- Supplemental Oxygen
- Lung Transplant
- Anti-inflammatory or immunosuppressive agents for colitis

Reference

OMIM Bookshelf

syndrome, albinism