Albinism

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What is albinism?

- Albinism is a defect of melanin production that results in little or no color (pigment) in the skin, hair, and eyes
  - Melanin is a natural substance that gives color (pigment) to hair, skin, and the iris of the eye.
    - produced by cells in the skin called melanocytes
  - Melanin also helps protect the skin from the sun

Symptoms

- Absence of color in the hair, skin, or iris of the eye
- Lighter than normal skin and hair
- Patchy, missing skin color
- Crossed eyes (strabismus)
- Light sensitivity (photophobia)
- Rapid eye movements (nystagmus)
- Vision problems(usually

Types of Albinism

- Oculocutaneous albinism
  - Most severe form of albinism
- Ocular albinism
- Hermansky-Pudlak syndrome

Oculocutaneous Albinism

- Type 1 (OCA1)
  - White hair, very pale skin, light-colored irises
  - Result from changes in TYR gene
- Type 2 (OCA2)
  - Less severe than Type 1
  - Creamy white skin color and light yellow, blond, or light brown hair
  - Mutations in the (“P” gene)
- Type 3 (OCA3)
  - Rufous Oculocutaneous Albinism (affects dark- skinned people)
  - Reddish-brown skin, ginger or red hair, and hazel or brown irises
  - TYRP1 mutation
- Type 4(OCA4)
  - Symptoms similar to Type 2
  - Mutations in SLC45A2 gene

Oculocutaneous Albinism (cont.)

- Four types of oculocutaneous albinism follow autosomal recessive pattern
  - both copies of the gene in each cell have mutations
  - parents each carry one copy of the mutated gene, but do not show signs and symptoms of the condition

- Frequency
  - 1 in 20,000 born with condition
  - Types 1 and 2 most common
  - Types 3 and 4 less common
  - Type 2 occurs mostly in African Americans, Native Americans, and sub-Saharan Africans
  - Type 3 most common in southern Africa
  - Type 4 most common in Japanese and Korean populations

Ocular Albinism

- Mainly affects the eyes (reduces pigmentation in the iris)
- Most common form Nettleship-Falls/ Type 1
- Affects 1 in 60,000 males (less common in women)
  - X-linked pattern
  - Men only have only 1 X chromosome so one altered copy is sufficient to develop symptoms
- Mutation in the GPR143 gene
  - Responsible for making a protein that plays a role in the pigmentation of the eyes and skin; controls growth of melanosomes (which store melanin)

Hermansky-Pudlak syndrome

- Type of albinism that includes
  - bleeding tendency
  - lung disease (pulmonary fibrosis)
  - bowel disease
  - kidney disease
  - Issues with blood clotting
  - Abnormal platelets
- 8 types
  - 1 and 4 most severe
  - 1,2, and 4 only types associated with pulmonary fibrosis
  - 3,5, and 6 have mildest symptoms
  - Little information known about types 7 and 8

Hermansky-Pudlak syndrome (cont.)

- Autosomal recessive
- 8 genes associated with disorder
  - Affects genes used to signal the making of 4 protein complexes
  - Complexes involved in formation of lysosome-related organelles
  - Mutations in associated genes prevent formation of LROs/ impair function of cell structures
    - Absence of LROs cause bleeding problems
    - Displays of albinism because LROs within melanocytes can't produce or distribute substance that gives hair, skin, and eyes their color

Treatment

- Treatment aimed to relieve symptoms
  - Visual Rehabilitation
    - Glasses often recommended to correct vision/ eye position
    - Eye muscle surgery to correct nystagmus
    - For strabismus some doctors recommend using eye patches
  - Advised to avoid the sun
  - Advised to use sunscreen /cover up completely

Works Consulted

- http://www.nlm.nih.gov/medlineplus/ency/article/002256.htm
- http://www.albinism.org/publications/hps.html
- http://www.medindia.net/patients/patientinfo/how-can-albinism-be-treated.htm
- http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002450/