Albinism

Main Features

- Oculocutaneous Albinism (OCA): Hypopigmentation of skin and hair with characteristic eye findings
- Ocular Albinism (OA): Normal skin and hair pigmentation with characteristic eye findings

Eye Findings

- Refractive error: significant myopia, hyperopia and/or astigmatism
- Motility: Nystagmus, esotropia
- Vision: Variable 20/40-20/200
- Iris: Transillumination defects (from complete lack of pigmentation in OCA1 to variable small defects in OA)
- Fundus: Hypopigmentation, foveal hypoplasia
- Optic nerve function: Abnormal visual evoked potentials (representing abnormally high amount of decussation of ganglion cell fibers)

Etiology

- OCA: autosomal recessive
  - OCA1A (commonly recognized type) occurs in 1/40,000 with 1/100 are carriers
    - Two TYR gene mutations (11q14-q21) causing no tyrosinase to be produced; no melanin
  - OCA1B: Two TYR gene mutations causing at least one copy of a partially active tyrosinase enzyme: less melanin
  - OCA2: P gene mutation (15q11.2-q12) hair pigmented but not skin, some iris pigment
  - OCA3: TYRP1 gene mutation (9p23) described in those of african descent
  - OCA4: MAPT gene mutation (SLC45A2)
  - Hermansky-Pudlak syndrome: mutation in any of HPS1 (10q23.1), HPS2 (5q13) , HPS3 (3q24), HPS4 (22q11.2-q12.2), HPS5-9
  - Chediak-Higashi syndrome: chromosome 1
- OA: X-linked
  - OA1 gene mutations (Xp22): males with normal hair and skin pigment, abnormal melanosome production

Other Findings

- OCA1A: No pigment of hair or skin, coarse rough skin, unpigmented nevi, solar

keratoses (premalignant) basal cell or squamous cell carcinomas (actually quite rare due to good prevention), skin melanocytes are present but melanoma rare
- OCA1B: white to very light hair and skin at birth with variable amounts of darkening, pigmented nevi and freckles can develop
- OCA2: Pigmented hair at birth, no generalized skin pigmentation but pigmented nevi and freckles can develop
- OCA4: Variable pigmentation of skin and hair, similar to OCA2
- Hermansky-Pudlak syndrome
    - Potentially lethal subtype
    - Bleeding diathesis
    - Pulmonary Fibrosis
    - Granulomatous colitis
- Chediak-Higashi syndrome
    - Potentially lethal subtype
    - Congenital Immunodeficiency causing infections of skin and respiratory tract
    - Bleeding Diathesis
    - Progressive Neurodegeneration
- OA1: giant melanosomes on skin biopsy if preformed, defect is in melanosome production, can have late onset sensorineural deafness, female carriers have mosaic pigmentation of peripheral fundus
- OA2: Families from Aland Islands in the Sea of Bothnia, female carriers do not have a mosaic pattern, possibly a type of CSNB (310500), (Xp11.4-p11.23) OMIM: 300600,
- OA3: ocular albinism that is autosomal recessive: OMIM: 203310 (6q13-q15) or (15q11.2-q12)
- OA with sensorineural deafness: OMIM: 103470 (11q14-q21, 3p14.1-p12.3)
    - Waardenberg Syndrome type II with ocular albinism, mutation in the transcription factor MITF which regulates the TYR gene

References

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- OCA1 GeneReview
- OCA2 GeneReview
- OCA4 GeneReview
- X-linked OA GeneReview

syndrome, albinism