Waardenburg Syndrome

Main Features

- Congenital sensorineural hearing loss
- White forelock
- Hair hypopigmentation
- Iris pigmentary abnormality

Eye Findings

- Iris pigmentary abnormality
  - Complete heterochromia
  - Segmental heterochromia
  - Complete hypoplastic blue iridies
- Telecanthus
- Choroidal hypopigmentation

Other Findings

- Skin hypopigmentation
- Synophrys/medial eyebrow flare
- Nasal anomalies
  - broad high nasal root
  - prominent columella
  - hypoplastic nasal alae
- Premature gray hair (age <30 years)
- Hirschsprung disease
- Cleft lip and palate
- Spina bifida

Etiology

- Autosomal Dominant
- Mutations in PAX3, MITF, SNAI2, SOX10, EDN3, EDNRB genees

Resources

- Waardenburg syndrome: iris and choroidal hypopigmentation, findings on anterior and posterior segment imaging. Shields CL et al. JAMA Ophthalmol. 2013;131(9):1167-1173
- Waardenburg Syndrome in Medline Plus

syndrome