

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 genes

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

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