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Spot diagnosis 1





Waardenburg syndrome

- Genetic disorders affecting neural crest cell development
- Most commonly inheritance- AD
- Clinical manifestation
 1. Pigmentary abnormalities
 - Heterochromia iridis or hypopigmented irides, White forelock
 - Patchy skin depigmentation
 2. Craniofacial features
 - Broad nasal root
 - Dystopia canthorum
 3. Congenital SNHL



Waardenburg syndrome

Clinical subtypes

- Type 1: *PAX3* mutation; dystopia canthorum and hearing loss
- Type 2: *MITF* mutation; similar features without dystopia canthorum
- Type 3 (Klein-Waardenburg): *PAX3* mutation with musculoskeletal abnormalities
- Type 4 (Waardenburg-Shah): Associated with Hirschsprung disease
 - **Some *SOX10* mutations create overlap with congenital hypogonadotropic hypogonadism (CHH)**
 - Possible association with Kallmann syndrome
 - May present with delayed puberty and GnRH deficiency

Molecular genetic

- *PAX3*, *MITF*, *SOX-10* mutation



Waardenburg syndrome

Management

- Replace deficient sex hormone
- Early identification of hearing impairment is critical
- Treatment options:
 - Hearing aids
 - Cochlear implantation
- Sun protection for hypopigmented skin
- Genetic counseling