

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** Waardenburg Syndrome
- **Test Code:** 3182

Related Diseases & Genes:

- Waardenburg syndrome, type 1 (OMIM #: 193500) || Gene: PAX3 (OMIM #: 606597)

Synonyms:

- WAARDENBURG SYNDROME WITH DYSTOPIA CANTHORUM
- PAX3 Mutations

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2-3 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dysmorphology

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