

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** FOXL2 Mutation
- **Test Code:** 2167

Related Diseases & Genes:

- Blepharophimosis, epicanthus inversus, and ptosis, type 1 (OMIM #: 110100) || Gene: FOXL2 (OMIM #: 605597)
- Blepharophimosis, epicanthus inversus, and ptosis, type 2 (OMIM #: 110100)
- Premature ovarian failure 3 (OMIM #: 608996)

Synonyms:

- FORKHEAD TRANSCRIPTION FACTOR FOXL2
- PITUITARY FORKHEAD FACTOR, MOUSE, HOMOLOG OF; PFRK

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 7-10 days

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dysmorphology

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