

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *TUBB3* Gene Sequence Analysis
- **Test Code:** 3493

Related Diseases & Genes:

- “Cortical dysplasia, complex, with other brain malformations” (OMIM #: 614039) || Gene: *TUBB3* (OMIM #: 602661)
- “Fibrosis of extraocular muscles, congenital, 3A” (OMIM #: 600638)

Synonyms:

- “TUBULIN, BETA-3”
- TUBB4
- “TUBULIN, BETA, CLASS III”

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
- Ophthalmogenetics
- Neurogenetics

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