

## GENETIC DISEASES EVALUATION CENTER

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### Test Name & Code:

- **Test Name:** TERT Sequencing
- **Test Code:** 4830

### Related Diseases & Genes:

- “Dyskeratosis congenita, autosomal dominant 2” (OMIM #: 613989) || Gene: *TERT* (OMIM #: 187270)
- “Dyskeratosis congenita, autosomal recessive 4” (OMIM #: 613989)
- “Leukemia, acute myeloid” (OMIM #: 601626)
- “Melanoma, cutaneous malignant, 9” (OMIM #: 615134)
- “Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1” (OMIM #: 614742)

### Synonyms:

- TELOMERASE CATALYTIC SUBUNIT
- TCS1
- EST2

### Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 weeks

### 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):

- Multisystem Diseases

