

GENETIC DISEASES EVALUATION CENTER

duzengenetics.com • duzen.com.tr

Test Name & Code:

- **Test Name:** Carnitine Palmitoyltransferase II Deficiency
- **Test Code:** 4788

Related Diseases & Genes:

- “CPT deficiency, hepatic, type II” (OMIM #: 600649) || Gene: CPT2 (OMIM #: 600650)
- “CPT II deficiency, lethal neonatal” (OMIM #: 608836)
- Myopathy due to CPT II deficiency (OMIM #: 255110)
- “Encephalopathy, acute, infection-induced, 4, susceptibility to” (OMIM #: 614212)

Synonyms:

- CARNITINE PALMITOYLTRANSFERASE II
- CPT II

Test Details:

- **Methodology:** Next Generation DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 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):

- Inborn Errors of Metabolism

+90 (312) 468-7010 • +90 (212) 272-4800 genetics@duzen.com.tr

duzengenetics.com • duzen.com.tr

For yesterday, today and *tomorrows...*

