

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** AR Deletion/Duplication Analysis
- **Test Code:** 4822

Related Diseases & Genes:

- Androgen insensitivity (OMIM #: 300068) || Gene: AR (OMIM #: 313700)
- “Androgen insensitivity, partial, with or without breast cancer” (OMIM #: 312300)
- “Hypospadias 1, X-linked” (OMIM #: 300633)
- Spinal and bulbar muscular atrophy of Kennedy (OMIM #: 313200)
- “Prostate cancer, susceptibility to” (OMIM #: 176807)

Synonyms:

- ANDROGEN RECEPTOR
- DIHYDROTESTOSTERONE RECEPTOR
- DHTR
- “NUCLEAR RECEPTOR SUBFAMILY 3, GROUP C, MEMBER 4”
- NR3C4

Test Details:


- **Methodology:** MLPA
- **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):

- Dysmorphology
- Reproductive Genetics
- Inborn Errors of Metabolism

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

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For yesterday, today and *tomorrows...*

