

GENETIC DISEASES EVALUATION CENTER

duzengenetics.com • duzen.com.tr

Test Name & Code:

- **Test Name:** SOX9 Mutations
- **Test Code:** 4030

Related Diseases & Genes:

- Acampomelic campomelic dysplasia (OMIM #: 114290) || Gene: SOX9 (OMIM #: 608160)
- Campomelic dysplasia (OMIM #: 114290)
- Campomelic dysplasia with autosomal sex reversal (OMIM #: 114290)

Synonyms:

- SRY-RELATED HMG-BOX GENE 9
- SRY-BOX 9
- XY sex reverse
- XY female

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** 5-10 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dysmorphology

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

duzengenetics.com • duzen.com.tr

For yesterday, today and *tomorrows...*

