Mutation	Amino acid change	Exon	Phenotype	HGMD accession	Reference/database
c.240G>C	p.Arg80Ser	Exon1	46,XY gonadal dysgenesis, complete	NA	PMID: 31745530
c.251A>G	p.Tyr84Cys	Exon1	46,XY sex reversal	CM172997	PMID: 28295047
c.331A>G	p.Arg111Gly	Exon1	46,XY sex reversal	NA	PMID:26005864
c.332G>T	p.Arg111Met	Exon1	46,XY sex reversal, type 4	NA	Clinvar
c.416T>A	p.Leu139Gln	Exon2	46,XY gonadal dysgenesis, complete	CM173500	PMID:27711951
c.884C>T	p.Pro295Leu	Exon4	46,XY sex reversal	CM990487	PMID:10332030
3'UTR +11insT	NA	NA	46,XYgonadal dysgenesis	CT109412	PMID:21340164
103Kb deletion of DMRT1	NA	Exons 1 and 2	46,XY gonadal dysgenesis	NA	PMID:20685758
35Kb deletion of DMRT1	NA	Exons 3 and 4	46,XY ovotesticular disorder of sexual development	CG125437	PMID:22573722
c.132C>T	p.Gly44Gly	Exon1	Meiotic arrest	NA	PMID:24934491
c.671A>G	p.Asn224Ser	Exon3	Infertility, SCO-syndrome	CM149515	PMID:24934491
c.783C>T	p.Pro261Pro	Exon3	Mixture of tubuli with varying stages of spermatogenesis	NA	PMID:24934491

c.991G>C	p.Asp331His	Exon5	SCO-syndrome；mixture of tubuli with varying stages of spermatogenesis	NA	PMID:24934491
c.-223_-219delCGAAAinsT	NA	NA	Azoospermia, non-obstructive	NA	PMID:26139570
141Kb deletion of DMRT1	NA	Exons 3,4and5	Azoospermia	CG135485	PMID:23555275
150Kb deletion of DMRT1	NA	Exons 3,4and5	Azoospermia	CG135484	PMID:23555275
55Kb deletion of DMRT1	NA	Exons 3 and 4	Azoospermia	CG135486	PMID:23555275

NA: not available; DSD: disorders of sex development; SCO-syndrome: complete bilateral Sertoli cell-only (SCO) syndrome