Homo sapiens sex determining region Y (SRY), RefSeqGene on chromosome Y

NCBI Reference Sequence: NG_011751.1

FASTA Graphics

Go to:

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REFERENCE   1  (bases 1 to 7897)
  AUTHORS   Calvo SE, Pagliarini DJ and Mootha VK.
  TITLE     Upstream open reading frames cause widespread reduction of protein
            expression and are polymorphic among humans
  JOURNAL   Proc Natl Acad Sci U S A 106 (18), 7507-7512 (2009)
   PUBMED   19372376
REFERENCE   2  (bases 1 to 7897)
  AUTHORS   Sekido R and Lovell-Badge R.
  TITLE     Sex determination and SRY: down to a wink and a nudge?
  JOURNAL   Trends Genet 25 (1), 19-29 (2009)
   PUBMED   19027189
  REMARK    Review article
REFERENCE   3  (bases 1 to 7897)
  AUTHORS   Clepet C, Schafer AJ, Sinclair AH, Palmer MS, Lovell-Badge R and
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  TITLE     The human SRY transcript
  JOURNAL   Hum Mol Genet 2 (12), 2007-2012 (1993)
   PUBMED   8111368
REFERENCE   4  (bases 1 to 7897)
  AUTHORS   Behlke MA, Bogan JS, Beer-Romero P and Page DC.
  TITLE     Evidence that the SRY protein is encoded by a single exon on the
            human Y chromosome
  JOURNAL   Genomics 17 (3), 736-739 (1993)
   PUBMED   8244390
REFERENCE   5  (bases 1 to 7897)
  AUTHORS   Su H and Lau YF.
  TITLE     Identification of the transcriptional unit, structural
            organization, and promoter sequence of the human sex-determining
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JOURNAL Am J Hum Genet 52 (1), 24-38 (1993)
PUBMED 8434602

REFERENCE 6 (bases 1 to 7897)
AUTHORS Mohnach,L., Fechner,P.Y. and Keegan,C.E.
TITLE Nonsyndromic Disorders of Testicular Development
JOURNAL (in) Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Mirzaa G and Amemiya A (Eds.);
GENEREVIEWS((R));
(1993)
PUBMED 20301714

REFERENCE 7 (bases 1 to 7897)
AUTHORS Delot,E.C. and Vilain,E.J.
TITLE Nonsyndromic 46,XX Testicular Disorders of Sex Development
JOURNAL (in) Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Mirzaa G and Amemiya A (Eds.);
GENEREVIEWS((R));
(1993)
PUBMED 20301589

REFERENCE 8 (bases 1 to 7897)
AUTHORS Sinclair AH, Berta P, Palmer MS, Hawkins JR, Griffiths BL, Smith MJ, Foster JW, Frischauf AM, Lovell-Badge R and Goodfellow PN.
TITLE A gene from the human sex-determining region encodes a protein with homology to a conserved DNA-binding motif
JOURNAL Nature 346 (6281), 240-244 (1990)
PUBMED 1695712

COMMENT REVIEWED REFSEQ: This record has been curated by NCBI staff. The reference sequence was derived from AC006040.3.
This sequence is a reference standard in the RefSeqGene project.

Summary: This intronless gene encodes a transcription factor that is a member of the high mobility group (HMG)-box family of DNA-binding proteins. This protein is the testis-determining factor (TDF), which initiates male sex determination. Mutations in this gene give rise to XY females with gonadal dysgenesis (Swyer syndrome); translocation of part of the Y chromosome containing this gene to the X chromosome causes XX male syndrome. [provided by RefSeq, Jul 2008].

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