DISEASE:
46,XX testicular difference of sex development

NAME:	46,XX testicular difference of sex development
DESCRIPTION:	A rare disorder of sex development (DSD) associated with a 46, XX karyotype and characterized by male external genitalia, ranging from normal to atypical with associated testosterone deficiency.
ORPHACODE:	393
SYNONYMS:	46,XX testicular DSD 46,XX testicular disorder of sex development De la Chapelle syndrome XX, male syndrome
XREF(S):	Orphanet MeSH OMIM OMIM OMIM ICD-10

ANALYTE(S):	SOX9 SRY SOX3 NR0B1 NR5A1
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RELATED CONTENT

Related Genetic Tests

- Hypogonadotropic hypogonadism (33 genes)
- Premature Ovarian Failure/Primary Ovarian Insufficiency (POF/POI) (32 genes)
- Sex determining region Y
- Sex determining region Y

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- nuclear receptor subfamily 0 group B member 1
- nuclear receptor subfamily 5 group A member 1
- SRY-box transcription factor 3
- SRY-box transcription factor 9
- sex determining region Y

Related Gene Panels

- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG

- Hypogonadotropic hypogonadism (33 genes) - VUB
- Premature Ovarian Failure/Insufficiency (32 genes) - VUB
