DISEASE:
46,XY complete gonadal dysgenesis

NAME:	46,XY complete gonadal dysgenesis
DESCRIPTION:	A rare disorder/difference of sex development (DSD) associated with absence in gonadal development that results in the presence of female appearing external and internal genitalia in presence of a 46,XY karyotype.
ORPHACODE:	242
SYNONYMS:	46,XY CGD 46,XY pure gonadal dysgenesis Swyer syndrome
XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	DHX37 WT1 SOX9 SRY DHH NR5A1 NR0B1 CBX2 MAP3K1 DMRT1
CREATED:	13 May 2019 - 01:02
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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

- chromobox 2
- desert hedgehog signaling molecule
- DEAH-box helicase 37
- doublesex and mab-3 related transcription factor 1
- mitogen-activated protein kinase kinase kinase 1
- nuclear receptor subfamily 0 group B member 1
- nuclear receptor subfamily 5 group A member 1
- SRY-box transcription factor 9
- sex determining region Y
- WT1 transcription factor

Related Gene Panels

- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
- Hypogonadotropic hypogonadism (33 genes) - VUB
- Premature Ovarian Failure/Insufficiency (32 genes) - VUB
