DISEASE:
46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency

NAME:	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
DESCRIPTION:	A rare difference of sex development characterized by 17-beta hydroxysteroid dehydrogenase 3 deficiency that affects individuals with a 46,XY karyotype leading to underandrogenization of the genitalia.
ORPHACODE:	752
SYNONYMS:	17-beta-hydroxysteroid dehydrogenase 3 deficiency 17-ketoreductase deficiency 17-ketosteroidreductase deficiency 46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	HSD17B3
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- hydroxysteroid 17-beta dehydrogenase 3