DISEASE:
46,XX gonadal dysgenesis

NAME:	46,XX gonadal dysgenesis
DESCRIPTION:	A rare disorder/difference of sex development characterized by a primary ovarian defect, either a failure of the gonads to develop or resistance to gonadotrophin stimulation which leads to premature ovarian failure (POF) in otherwise phenotypically female 46,XX individuals.
ORPHACODE:	243
SYNONYMS:	46,XX complete gonadal dysgenesis 46,XX ovarian dysgenesis 46,XX pure gonadal dysgenesis Hypergonadotropic ovarian dysgenesis XX female gonadal dysgenesis XX-GD

XREF(S):	Orphanet OMIM MeSH ICD-10 OMIM OMIM OMIM OMIM OMIM
ANALYTE(S):	NR5A1 MRPS22 FSHR BMP15 PSMC3IP NUP107 BNC1 ZSWIM7 POLR3H MSH4 SPIDR
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RELATED CONTENT

Related Genetic Tests

- Premature Ovarian Failure/Primary Ovarian Insufficiency (POF/POI) (32 genes)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- bone morphogenetic protein 15
- basonuclin zinc finger protein 1
- follicle stimulating hormone receptor
- mitochondrial ribosomal protein S22
- mutS homolog 4
- nuclear receptor subfamily 5 group A member 1
- nucleoporin 107
- RNA polymerase III subunit H
- PSMC3 interacting protein
- scaffold protein involved in DNA repair
- zinc finger SWIM-type containing 7

Related Gene Panels

- Premature Ovarian Failure/Insufficiency (32 genes) - VUB
