DISEASE:
Perrault syndrome

NAME:	Perrault syndrome
DESCRIPTION:	Perrault syndrome (PS) is characterized by the association of ovarian dysgenesis in females with sensorineural hearing impairment. In more recent PS reports, some authors have described neurologic abnormalities, notably progressive cerebellar ataxia and intellectual deficit.
ORPHACODE:	2855
SYNONYMS:	XX gonodal dysgenesis-deafness syndrome XX gonodal dysgenesis-hearing loss syndrome
XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM ICD-10 OMIM

ANALYTE(S):	ERAL1 HSD17B4 TWNK HARS2 CLPP LARS2
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Perrault syndrome (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- caseinolytic mitochondrial matrix peptidase proteolytic subunit
- Era like 12S mitochondrial rRNA chaperone 1
- histidyl-tRNA synthetase 2, mitochondrial
- hydroxysteroid 17-beta dehydrogenase 4
- leucyl-tRNA synthetase 2, mitochondrial
- twinkle mtDNA helicase

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Perrault syndrome (5 genes) - UZA

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