DISEASE:
Primary ciliary dyskinesia-retinitis pigmentosa syndrome

NAME:	Primary ciliary dyskinesia-retinitis pigmentosa syndrome
DESCRIPTION:	Primary ciliary dyskinesia - retinitis pigmentosa is an X-linked ciliary dysfunction of both respiratory epithelium and photoreceptors of the retina leading to ocular disorders (mild night blindness, constriction of the visual field, and scotopic and photopic ERG responses reduced to 30-60%) associated with primary ciliary dyskinesia (see this term) manifestations (chronic bronchorrhea with bronchoectasis and chronic sinusitis) and sensorineural hearing loss.
ORPHACODE:	247522
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	RPGR
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- Primary Ciliary Dyskinesia (gene panel)

Related Laboratories

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

Related Analytes

- retinitis pigmentosa GTPase regulator

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- Primary Ciliary Dyskinesia (61 genes) - KUL