DISEASE:
Primary ciliary dyskinesia

NAME:	Primary ciliary dyskinesia
DESCRIPTION:	A rare, genetically heterogeneous, primarily respiratory disorder characterized by chronic upper and lower respiratory tract disease. Approximately half of the patients have an organ laterality defect (situs inversus totalis or situs ambiguus/heterotaxy).
ORPHACODE:	244
SYNONYMS:	PCD

XREF(S):	Orphanet OMIM MedDRA ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	DNAH1 TTC12 NEK10 DNAAF6 CFAP221 SPEF2 DNAJB13 ODAD4 NME5 MCIDAS GAS2L2 OFD1 DNAH11 DNAH5 DNAI1 NME8 DNAI2 DNAAF2 RSPH9 RSPH4A DNAAF1 CCDC39 CCDC40 DNAL1 DNAAF3 CCDC103 DNAAF5 HYDIN DNAAF11 ODAD1 DRC1 ODAD2 DNAAF4 RSPH1 ZMYND10 CFAP298 CCDC65 SPAG1 CCNO

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

- coiled-coil domain containing 103
- coiled-coil domain containing 39
- coiled-coil domain containing 40
- coiled-coil domain containing 65
- cyclin O
- cilia and flagella associated protein 221
- cilia and flagella associated protein 298
- cilia and flagella associated protein 300
- cilia and flagella associated protein 74
- dynein axonemal assembly factor 1
- dynein axonemal assembly factor 11
- dynein axonemal assembly factor 2
- dynein axonemal assembly factor 3

- dynein axonemal assembly factor 4
- dynein axonemal assembly factor 5
- PIH1 domain containing 3
- dynein axonemal heavy chain 1
- dynein axonemal heavy chain 11
- dynein axonemal heavy chain 5
- dynein axonemal heavy chain 9
- dynein axonemal intermediate chain 1
- dynein axonemal intermediate chain 2
- DnaJ heat shock protein family (Hsp40) member B13
- dynein axonemal light chain 1
- dynein regulatory complex subunit 1
- forkhead box J1
- growth arrest specific 2 like 2
- growth arrest specific 8
- HYDIN axonemal central pair apparatus protein
- leucine rich repeat containing 56
- multiciliate differentiation and DNA synthesis associated cell cycle protein
- NIMA related kinase 10
- NME/NM23 family member 5
- NME/NM23 family member 8
- outer dynein arm docking complex subunit 1
- outer dynein arm docking complex subunit 2
- outer dynein arm docking complex subunit 3
- outer dynein arm docking complex subunit 4
- OFD1 centriole and centriolar satellite protein
- retinitis pigmentosa GTPase regulator
- radial spoke head component 1
- radial spoke head 3
- radial spoke head component 4A
- radial spoke head component 9
- sperm associated antigen 1
- sperm flagellar 2

- serine/threonine kinase 36
- tetratricopeptide repeat domain 12
- zinc finger MYND-type containing 10

Related Gene Panels

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