

GENETIC TEST:
Primary ciliary dyskinesia (PCD) Heterotaxyies (gene panel)

FULL NAME:	Primary ciliary dyskinesia (PCD) Heterotaxyies (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, DNA
METHOD CATEGORY:	Sequence analysis: entire coding region Whole Exome Sequencing (WES)
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-09-11 / 2026-09-10
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	15 Dec 2020 - 16:20
CHANGED:	08 Jul 2022 - 10:27

URL:	https://www.cmgg.be/assets/bestanden/Genpanel-Heterotaxie-PCD.pdf
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- centrosomal protein 290
- cilia and flagella associated protein 298
- cilia and flagella associated protein 300
- cilia and flagella associated protein 45
- cilia and flagella associated protein 52
- cilia and flagella associated protein 53
- cilia and flagella associated protein 58
- cripto, FRL-1, cryptic family 1
- CF transmembrane conductance regulator
- chromodomain helicase DNA binding protein 7
- ciliated left-right organizer metalloproteinase
- Cbp/p300 interacting transactivator with Glu/Asp rich carboxy-terminal domain 2
- cysteine rich with EGF like domains 1
- dynein assembly factor with WD repeats 1
- diacylglycerol O-acyltransferase 2 like 6
- 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 12
- dynein axonemal heavy chain 14
- dynein axonemal heavy chain 17
- dynein axonemal heavy chain 5
- dynein axonemal heavy chain 8
- dynein axonemal heavy chain 9
- dynein axonemal intermediate chain 1
- dynein axonemal intermediate chain 2
- DnaJ heat shock protein family (Hsp40) member B13

- dynein regulatory complex subunit 1
- dynein cytoplasmic 1 heavy chain 1
- dynein light chain Tctex-type 2B
- elastin
- EvC ciliary complex subunit 1
- EvC ciliary complex subunit 2
- forkhead box F1
- forkhead box H1
- forkhead box J1
- growth arrest specific 2 like 2
- growth arrest specific 8
- GATA binding protein 4
- GATA binding protein 6
- growth differentiation factor 1
- gap junction protein alpha 1
- glypican 3
- heart and neural crest derivatives expressed 1
- hes family bHLH transcription factor 7
- HYDIN axonemal central pair apparatus protein
- intraflagellar transport 46
- inversin
- jagged canonical Notch ligand 1
- kinesin family member 7
- left-right determination factor 1
- left-right determination factor 2
- LMBR1 domain containing 1
- lamin A/C
- leucine rich repeat containing 56
- leucine zipper transcription factor like 1
- mitogen-activated protein kinase kinase 1
- mitogen-activated protein kinase kinase 2
- multiciliate differentiation and DNA synthesis associated cell cycle protein
- mediator complex subunit 13L

- multiple EGF like domains 8
- Meis homeobox 2
- MKS transition zone complex subunit 1
- matrix metalloproteinase 21
- meiosis specific nuclear structural 1
- MRE11 homolog, double strand break repair nuclease
- myosin heavy chain 6
- N-acetyltransferase 10
- NIMA related kinase 10
- NIMA related kinase 9
- NK2 homeobox 5
- NK2 homeobox 6
- NME/NM23 family member 5
- NME/NM23 family member 8
- nodal growth differentiation factor
- notch receptor 1
- notch receptor 2
- nephrocystin 3
- nuclear receptor subfamily 2 group F member 2
- nuclear receptor binding SET domain protein 1
- 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
- pitrilysin metalloproteinase 1
- polycystin 1 like 1, transient receptor potential channel interacting
- polycystin 2, transient receptor potential cation channel
- polyglutamine binding protein 1
- paired related homeobox 1
- protein tyrosine phosphatase non-receptor type 11
- Raf-1 proto-oncogene, serine/threonine kinase
- Ras like without CAAX 1

- [retinitis pigmentosa GTPase regulator](#)
- [radial spoke head component 1](#)
- [radial spoke head 3](#)
- [radial spoke head component 4A](#)
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- [serpin family E member 2](#)
- [SHOC2 leucine rich repeat scaffold protein](#)
- [shroom family member 3](#)
- [SMAD family member 2](#)
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- [SOS Ras/Rac guanine nucleotide exchange factor 1](#)
- [sperm associated antigen 1](#)
- [sperm flagellar 2](#)
- [serine/threonine kinase 36](#)
- [T-box transcription factor 1](#)
- [T-box transcription factor 5](#)
- [tectonic family member 2](#)
- [tumor protein p73](#)
- [tetratricopeptide repeat domain 12](#)
- [tetratricopeptide repeat domain 8](#)
- [ubiquitin protein ligase E3 component n-recognin 1](#)
- [WD repeat domain 35](#)
- [WD repeat domain 47](#)
- [zinc finger protein, FOG family member 2](#)
- [Zic family member 3](#)
- [zinc metalloproteinase STE24](#)
- [zinc finger MYND-type containing 10](#)
- [zinc finger protein 423](#)

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