GENETIC TEST:
Ciliopathy (gene panel)

FULL NAME:	Ciliopathy (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
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:	29 Jul 2019 - 12:55
CHANGED:	13 Dec 2022 - 09:07
URL:	https://www.cmgg.be/assets/bestanden/Genpanel-Ciliopathie-v4.pdf

RELATED CONTENT

Related Diseases

- Achromatopsia
- Acrocallosal syndrome
- Acrofacial dysostosis, Weyers type
- Ataxia-telangiectasia-like disorder
- Autosomal dominant polycystic kidney disease
- Autosomal recessive polycystic kidney disease
- Bardet-Biedl syndrome
- Caroli disease
- Cone rod dystrophy
- Cranioectodermal dysplasia
- Ellis Van Creveld syndrome
- Heart defect-tongue hamartoma-polysyndactyly syndrome
- Hydrolethalus
- Infantile nephronophthisis
- Isolated neonatal sclerosing cholangitis
- Jeune syndrome
- Joubert syndrome
- Joubert syndrome with Jeune asphyxiating thoracic dystrophy
- Joubert syndrome with hepatic defect
- Joubert syndrome with oculorenal defect
- Joubert syndrome with renal defect
- Juvenile nephronophthisis
- Late-onset nephronophthisis
- Leber congenital amaurosis
- MORM syndrome
- McKusick-Kaufman syndrome

- Meckel syndrome
- Multiple epiphyseal dysplasia, Al-Gazali type
- Non-syndromic male infertility due to sperm motility disorder
- Orofaciodigital syndrome type 1
- Orofaciodigital syndrome type 2
- Orofaciodigital syndrome type 3
- Orofaciodigital syndrome type 6
- Primary ciliary dyskinesia
- Primary ciliary dyskinesia-retinitis pigmentosa syndrome
- Renal-hepatic-pancreatic dysplasia
- Retinitis pigmentosa
- Saldino-Mainzer syndrome
- Senior-Boichis syndrome
- Senior-Loken syndrome
- Short rib-polydactyly syndrome type 5
- Short rib-polydactyly syndrome, Beemer-Langer type
- Short rib-polydactyly syndrome, Majewski type
- Short rib-polydactyly syndrome, Saldino-Noonan type
- Short rib-polydactyly syndrome, Verma-Naumoff type
- X-linked recessive intellectual disability-macrocephaly-ciliary dysfunction syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- activin A receptor type 2B
- ADAM metallopeptidase with thrombospondin type 1 motif 9
- Abelson helper integration site 1

- ALMS1 centrosome and basal body associated protein
- ankyrin repeat and sterile alpha motif domain containing 6
- ADP ribosylation factor like GTPase 13B
- ADP ribosylation factor like GTPase 3
- ADP ribosylation factor like GTPase 6
- armadillo repeat containing 9
- B9 domain containing 1
- B9 domain containing 2
- BBSome interacting protein 1
- Bardet-Biedl syndrome 1
- Bardet-Biedl syndrome 10
- Bardet-Biedl syndrome 12
- Bardet-Biedl syndrome 2
- Bardet-Biedl syndrome 4
- Bardet-Biedl syndrome 5
- Bardet-Biedl syndrome 7
- Bardet-Biedl syndrome 9
- C2 domain containing 3 centriole elongation regulator
- chibby family member 1, beta catenin antagonist
- coiled-coil and C2 domain containing 2A
- coiled-coil domain containing 103
- coiledcoiled-coil domain containing 172-coil domain containing 172
- coiled-coil domain containing 28B
- coiled-coil domain containing 32
- coiled-coil domain containing 39
- coiled-coil domain containing 40
- coiled-coil domain containing 65
- coiled-coil domain containing 96
- cyclin O
- centromere protein F
- centrosomal protein 104
- centrosomal protein 120
- centrosomal protein 164

- centrosomal protein 290
- centrosomal protein 295
- centrosomal protein 41
- centrosomal protein 55
- centrosomal protein 83
- cilia and flagella associated protein 251
- cilia and flagella associated protein 298
- cilia and flagella associated protein 300
- cilia and flagella associated protein 410
- cilia and flagella associated protein 418
- cilia and flagella associated protein 44
- cilia and flagella associated protein 53
- cilia and flagella associated protein 69
- cripto, FRL-1, cryptic family 1
- ciliogenesis associated kinase 1
- ciliogenesis and planar polarity effector complex subunit 1
- crumbs cell polarity complex component 2
- centrosome and spindle pole associated protein 1
- doublecortin domain containing 2
- DEAD-box helicase 59
- deuterosome assembly protein 1
- 7-dehydrocholesterol reductase
- discs large MAGUK scaffold protein 5
- 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 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 2 heavy chain 1
- dynein 2 intermediate chain 1
- dynein 2 intermediate chain 2
- dynein cytoplasmic 2 light intermediate chain 1
- dynein light chain Tctex-type 2B
- EvC ciliary complex subunit 1
- EvC ciliary complex subunit 2
- exocyst complex component 3 like 2
- exocyst complex component 6B
- exocyst complex component 8
- exostosin like glycosyltransferase 3
- family with sequence similarity 149 member B1
- family with sequence similarity 166 member B
- forkhead box F1
- fuzzy planar cell polarity protein
- growth arrest specific 2 like 2
- growth arrest specific 8
- GLI family zinc finger 3
- GLIS family zinc finger 2
- HNF1 homeobox B
- HYDIN axonemal central pair apparatus protein
- HYLS1 centriolar and ciliogenesis associated
- intraflagellar transport 122
- intraflagellar transport 140
- intraflagellar transport 172
- intraflagellar transport 27
- intraflagellar transport 43

- intraflagellar transport 52
- intraflagellar transport 74
- intraflagellar transport 80
- intraflagellar transport 81
- inositol polyphosphate-5-phosphatase E
- integrator complex subunit 13
- inturned planar cell polarity protein
- inversin
- IQ motif containing B1
- IQ motif containing E
- katanin interacting protein
- potassium channel tetramerization domain containing 3
- KIAA0586
- KIAA0753
- kinesin family member 14
- kinesin family member 3B
- kinesin family member 7
- lamin B receptor
- lebercilin LCA5
- leucine rich repeat containing 34
- leucine rich repeat containing 45
- leucine rich repeat containing 56
- leucine zipper transcription factor like 1
- mitogen-activated protein kinase binding protein 1
- multiciliate differentiation and DNA synthesis associated cell cycle protein
- MKKS centrosomal shuttling protein
- MKS transition zone complex subunit 1
- matrix metallopeptidase 21
- MRE11 homolog, double strand break repair nuclease
- non-SMC condensin II complex subunit G2
- NIMA related kinase 1
- NIMA related kinase 9
- NME/NM23 family member 5

- NME/NM23 family member 8
- nephrocystin 1
- nephrocystin 3
- nephrocystin 4
- OCRL inositol polyphosphate-5-phosphatase
- 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
- phosphodiesterase 6D
- progesterone immunomodulatory binding factor 1
- phosphatidylinositol-4-phosphate 3-kinase catalytic subunit type 2 alpha
- polycystin 1, transient receptor potential channel interacting
- polycystin 2, transient receptor potential cation channel
- PKHD1 ciliary IPT domain containing fibrocystin/polyductin
- polyamine modulated factor 1 binding protein 1
- phosphomannomutase 2
- POC1 centriolar protein A
- POC1 centriolar protein B
- RAB28, member RAS oncogene family
- retinitis pigmentosa GTPase regulator
- RPGRIP1 like
- radial spoke head component 1
- radial spoke head 3
- radial spoke head component 4A
- radial spoke head component 9
- SBDS ribosome maturation factor
- sodium channel and clathrin linker 1
- sodium channel modifier 1
- SHH signaling and ciliogenesis regulator SDCCAG8
- solute carrier family 30 member 7
- sperm associated antigen 1

- serine/threonine kinase 36
- SUFU negative regulator of hedgehog signaling
- TBC1 domain family member 32
- tectonic family member 1
- tectonic family member 2
- tectonic family member 3
- transmembrane protein 107
- transmembrane protein 138
- transmembrane protein 17
- transmembrane protein 216
- transmembrane protein 218
- transmembrane protein 231
- transmembrane protein 237
- transmembrane protein 260
- transmembrane protein 67
- TOG array regulator of axonemal microtubules 1
- TOP1 binding arginine/serine rich protein, E3 ubiquitin ligase
- TRAF3 interacting protein 1
- tripartite motif containing 32
- tetratricopeptide repeat domain 21B
- tetratricopeptide repeat domain 23
- tetratricopeptide repeat domain 26
- tetratricopeptide repeat domain 6
- tetratricopeptide repeat domain 8
- tubulin gamma complex associated protein 4
- TUB like protein 1
- TUB like protein 3
- thioredoxin domain containing 15
- von Hippel-Lindau tumor suppressor
- vacuolar protein sorting 13 homolog B
- WD repeat containing planar cell polarity effector
- WD repeat domain 19
- WD repeat domain 35

- X-prolyl aminopeptidase 3
- zinc finger FYVE-type containing 19
- Zic family member 3
- zinc finger MYND-type containing 10
- zinc finger protein 423
- zinc finger SWIM-type containing 6

Related Gene Panels

- Ciliopathy (120 genes) - UGent
