Primary Ciliary Dyskinesia

Precision Panel

Overview

Primary Ciliary Dyskinesia (PCD) is a highly heterogeneous syndrome characterized by congenital impairment of mucociliary clearance (MCC). The underlying cause is a defect of cilia in the airways, making them unable to beat normally and move respiratory secretions. This defect also has an impact in sperm flagella, generating living but immotile spermatozoa and making patients infertile.

The most common defects causing this disease are found in any polypeptide within the axoneme of cilia, in proteins present in the ciliary membrane and matrix, or in proteins needed for the proper assembly of cilia. Depending on the component missing or defective, different clinical manifestations may develop, being the symptoms and disease onset dependent on the underlying genetic defect. Some mutations result in abnormal ultrastructure, while others cause abnormal function but preserved structure. Since nodal cilia are also defective in embryos, body asymmetry occurs randomly so that approximately 50 percent of the patients have situs inversus totalis. The mode of inheritance is mainly autosomal recessive.

The Igenomix Primary Ciliary Dyskinesia Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Primary Ciliary Dyskinesia Precision Panel is indicated for those patients with a clinical diagnosis or suspicion presenting with or without the following manifestations:

- Respiratory distress
- Rhinosinusitis
- Situs inversus
- Frequent otitis media
- Fatigue and headaches
- Decreased fertility or infertility

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of multidisciplinary treatment including pharmacological treatment in form of mucolytic agents and antibiotics to deal with frequent infections and exacerbations. Daily chest physiotherapy is commonly used to help reduce the microbial load. Surgical intervention in form of bilateral lung transplantation is also an option for patients with end-stage respiratory insufficiency.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

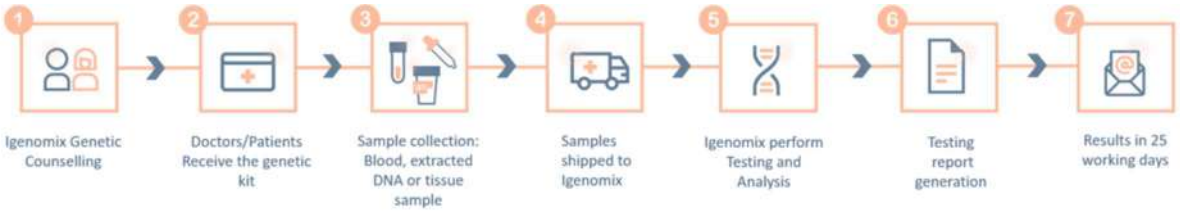
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	CLINVAR**	HGMD**
CCDC103	Primary Ciliary Dyskinesia	AR	99.92	4 of 4	6 of 6
CCDC39	Primary Ciliary Dyskinesia	AR	99.56	30 of 36	48 of 52
CCDC40	Primary Ciliary Dyskinesia	AR	98	35 of 35	50 of 50
CCDC65	Primary Ciliary Dyskinesia	AR	99.98	4 of 4	3 of 3
CCNO	Primary Ciliary Dyskinesia	AR	99.94	19 of 19	12 of 12
CENPF	Stromme Syndrome	AR	98.83	15 of 18	10 of 12
CFAP221	Primary Ciliary Dyskinesia	-	89.78	-	-
CFAP298	Primary Ciliary Dyskinesia	AR	-	-	-
CFAP300	Primary Ciliary Dyskinesia	AR	-	-	-
DNAAF1	Primary Ciliary Dyskinesia	AR	99.55	17 of 17	36 of 37
DNAAF11	Primary Ciliary Dyskinesia	AR	99.88	14 of 14	21 of 21
DNAAF2	Primary Ciliary Dyskinesia	AR	97.45	15 of 15	7 of 8
DNAAF3	Primary Ciliary Dyskinesia	AR	98.95	14 of 15	13 of 14
DNAAF4	Primary Ciliary Dyskinesia	AD,AR	99.27	-	-
DNAAF5	Primary Ciliary Dyskinesia	AR	89.27	-	-
DNAAF6	Primary Ciliary Dyskinesia	X,XR,G	99.63	-	-
DNAH1	Spermatogenic Failure, Primary Ciliary Dyskinesia	AR	100	17 of 17	58 of 58
DNAH11	Primary Ciliary Dyskinesia	AR	99.27	87 of 90	159 of 169
DNAH5	Primary Ciliary Dyskinesia, Situs Inversus	AR	100	188 of 188	277 of 278
DNAH8	Spermatogenic Failure	AR	99.75	24 of 25	12 of 12
DNAH9	Primary Ciliary Dyskinesia	AR	98.86	7 of 7	19 of 19
DNAI1	Kartagener Syndrome, Primary Ciliary Dyskinesia	AR	96.91	20 of 20	43 of 43
DNAI2	Primary Ciliary Dyskinesia, Situs Inversus	AR	98.89	25 of 25	8 of 8
DNAJB13	Primary Ciliary Dyskinesia	AR	99.94	2 of 2	3 of 3
DNAL1	Primary Ciliary Dyskinesia	AR	99.43	4 of 4	5 of 5
DRC1	Primary Ciliary Dyskinesia	AR	100	5 of 5	9 of 9
FOXJ1	Primary Ciliary Dyskinesia	AD	99.69	4 of 4	5 of 5
GAS2L2	Primary Ciliary Dyskinesia	AR	89	1 of 1	4 of 5

GAS8	Primary Ciliary Dyskinesia	AR	99.98	4 of 4	6 of 6
HYDIN	Primary Ciliary Dyskinesia	AR	81.7	3 of 6	45 of 63
INVS	Nephronophthisis, Senior-Loken Syndrome	AR	99.9	21 of 21	38 of 38
LRRC56	Primary Ciliary Dyskinesia	AR	99.77	4 of 4	5 of 5
MCIDAS	Primary Ciliary Dyskinesia	AR	99.92	6 of 6	4 of 4
NEK10	Primary Ciliary Dyskinesia	AR	99.95	4 of 4	3 of 3
NME8	Primary Ciliary Dyskinesia	AR	99.99	1 of 1	9 of 9
ODAD1	Primary Ciliary Dyskinesia	AR	99.68	7 of 9	10 of 10
ODAD2	Primary Ciliary Dyskinesia	AR	97.3	19 of 20	26 of 28
ODAD3	Primary Ciliary Dyskinesia	AR	95	10 of 10	4 of 4
ODAD4	Primary Ciliary Dyskinesia	AR	-	-	-
OFD1	Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	-	-
RPGR	Primary Ciliary Dyskinesia	X,XR,G	94	-	-
RSPH1	Primary Ciliary Dyskinesia	AR	100	14 of 14	10 of 10
RSPH3	Primary Ciliary Dyskinesia	AR	99.85	9 of 9	5 of 5
RSPH4A	Primary Ciliary Dyskinesia	AR	99.98	23 of 23	27 of 27
RSPH9	Primary Ciliary Dyskinesia	AR	100	9 of 9	13 of 13
SPAG1	Primary Ciliary Dyskinesia	AR	94.8	16 of 17	11 of 12
SPEF2	Spermatogenic Failure, Primary Ciliary Dyskinesia	AR	99.6	6 of 7	10 of 13
STK36	Primary Ciliary Dyskinesia	-	100	-	5 of 5
TTC12	Primary Ciliary Dyskinesia	AR	99.97	4 of 4	-
ZMYND10	Primary Ciliary Dyskinesia	AR	99.98	9 of 9	16 of 16

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant pathogenic and likely pathogenic variants, according to ClinVar and HGMD.

Methodology



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References

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