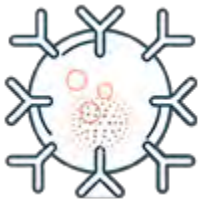
Inherited Disorders of the Complement System

Precision Panel



Overview

Inherited Disorders of the Complement System are rare disorders that predispose to bacterial infections and/or systemic lupus erythematosus (SLE). The complement system is a group of circulating proteins that participate in the innate immunity, bind to pathogens and destroys them. In addition to playing an important role in host defense against infection, the complement system is a mediator in both the pathogenesis and prevention of immune complex disease such as SLE. Inherited disorders of the complement system are associated with predictable defects in complement-dependent function, as the affected individual loses not only the activity of the deficient protein, but also the functions of the proteins in the cascade. Additionally, it can also be due to heterozygous deficiency. These are generally classified in two categories: 1) integral component defects and 2) regulatory component defects.

The Igenomix Inherited Disorders of the Complement System Precision Panel can be used for an accurate and directed diagnosis as well as differential diagnosis of recurrent infections 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 Inherited Disorders of the Complement System Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Sinopulmonary bacterial infections
- Bacteremia
- Meningitis
- Autoimmune disorders such as SLE

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 antimicrobial prophylaxis with antibacterial, antifungals, rapid recognition and treatment of infections as well as aggressive management of infectious complications.

For those patients presenting with autoimmune disease, treatment of these patients focuses on immunosuppressive therapy.

- 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)	HGMD**
C1QA	C1q Deficiency	AR	100	8 of 8
C1QB	C1q Deficiency	AR	99.91	8 of 8
C1QBP	Oxidative Phosphorylation Deficiency	AR	99.89	6 of 6
C1QC	C1q Deficiency	AR	100	11 of 11
C1S	Complement Component C1s Deficiency, Ehlers-Danlos Syndrome	AD	100	12 of 12
C2	Complement Component 2 Deficiency	AR	99.97	9 of 9
C3	Complement Component 3 Deficiency, Hemolytic Uremic Syndrome	AD,AR	100	123 of 124
C4A	Complement Component 4a Deficiency, Behçet Disease	AR	19.11	0 of 6
C4B	Complement Component 4b Deficiency	-	22.98	3 of 7
C5	C5 Deficiency	AR	99.98	19 of 19
C6	C6 Deficiency	AR	100	15 of 15
C7	C7 Deficiency	AR	99.63	28 of 30
C8A	Complement Component 8 Deficiency	AR	100	8 of 8
C8B	Complement Component 8 Deficiency	AR	100	11 of 11
C9	Complement Component 9 Deficiency	-	100	11 of 11
CCDC103	Ciliary Dyskinesia	AR	99.92	6 of 6
CCDC39	Ciliary Dyskinesia	AR	99.56	48 of 52
CCDC40	Ciliary Dyskinesia	AR	98	50 of 50
CCDC65	Ciliary Dyskinesia	AR	99.98	3 of 3
CCNO	Ciliary Dyskinesia	AR	99.94	12 of 12
CD46	Hemolytic Uremic Syndrome, Hellp Syndrome	AD,AR	100	83 of 84
CD55	Enteropathy	AR	98.21	5 of 6
CD59	Hemolytic Anemia, Immune-Mediated Polyneuropathy	AR	99.99	8 of 8
CFAP300	Ciliary Dyskinesia	AR	-	-
CFB	Complement Factor B Deficiency, Hemolytic Uremic Syndrome	AD,AR	100	26 of 26
CFD	Complement Factor D Deficiency	AR	99.88	3 of 3
CFH	Basal Laminar Drusen, Complement Factor H Deficiency, Hemolytic Uremic Syndrome, Macular Degeneration,Hellp Syndrome	AD,AR,MU,P	99.94	340 of 342
CFHR1	Hemolytic Uremic Syndrome, Macular Degeneration	AD,AR	88.29	0 of 9
CFHR3	Hemolytic Uremic Syndrome, Macular Degeneration	AD,AR	89.89	0 of 7
CFI	Complement Factor I Deficiency, Hemolytic Uremic Syndrome, Macular Degeneration, Hellp Syndrome	AD,AR	99.93	156 of 158
CFP	Properdin Deficiency	X,XR,G	100	-
COLEC11	Carnevale Syndrome, 3mc Syndrome	AR	100	11 of 11

CR2	Immunodeficiency	AD,AR	99.92	19 of 19
DGKE	Nephrotic Syndrome	AR	99.67	41 of 42
DNAAF1	Ciliary Dyskinesia	AR	99.55	36 of 37
DNAAF2	Ciliary Dyskinesia	AR	97.45	7 of 8
DNAAF3	Ciliary Dyskinesia	AR	98.95	13 of 14
DNAAF4	Ciliary Dyskinesia, Dyslexia	AD,AR	99.27	-
DNAAF5	Ciliary Dyskinesia	AR	89.27	-
DNAH11	Ciliary Dyskinesia	AR	99.27	159 of 169
DNAH5	Ciliary Dyskinesia	AR	100	277 of 278
DNAH9	Ciliary Dyskinesia	AR	98.86	19 of 19
DNAI1	Kartagener Syndrome, Ciliary Dyskinesia	AR	96.91	43 of 43
DNAI2	Ciliary Dyskinesia, Situs Inversus	AR	98.89	8 of 8
DNAL1	Ciliary Dyskinesia	AR	99.43	5 of 5
DNASE1L3	Lupus Erythematosus, Hypocomplementemic Urticarial Vasculitis	AR	100	4 of 4
DRC1	Ciliary Dyskinesia	AR	100	9 of 9
FCN3	Immunodeficiency	AR	99.98	1 of 1
GAS2L2	Ciliary Dyskinesia	AR	89	4 of 5
HYDIN	Ciliary Dyskinesia	AR	81.7	45 of 63
IRAK1	Lupus Erythematosus	-	96.2	-
LMNA	Cardiomyopathy, Charcot-Marie-Tooth Disease, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Mandibuloacral Dysplasia, Restrictive Dermopathy, Werner Syndrome, Hypergonadotropic Hypogonadism, Lipodystrophy	AD,AR	100	619 of 620
LMNB2	Barraquer-Simons Syndrome, Epilepsy, Lipodystrophy	AD,AR	95.03	5 of 5
LRRC6	Ciliary Dyskinesia	AR	99.88	21 of 21
MASP1	3mc Syndrome	AR	100	29 of 30
MASP2	Masp2 Deficiency	AR	99.68	7 of 7
MAT2A	Thoracic Aortic Aneurysm, Aortic Dissection	-	100	3 of 3
MCIDAS	Ciliary Dyskinesia	AR	99.92	4 of 4
NME8	Ciliary Dyskinesia	AR	99.99	9 of 9
ODAD1	Ciliary Dyskinesia	AR	99.68	10 of 10
ODAD2	Ciliary Dyskinesia	AR	97.3	26 of 28
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Ciliary Dyskinesia	X,XR,XD,G	98.09	-
PACS1	Mental Retardation, Intellectual Disability, Craniofacial Dysmorphism, Cryptorchidism	AD	97.98	3 of 3
PIGA	Hypotonia, Seizures, Paroxysmal Nocturnal Hemoglobinuria, West Syndrome	X,XR,G	97.98	-
RSPH1	Ciliary Dyskinesia	AR	100	10 of 10
RSPH4A	Ciliary Dyskinesia	AR	99.98	27 of 27
RSPH9	Ciliary Dyskinesia	AR	100	13 of 13
SERPING1	Angioedema, Complement Component 4 Deficiency	AD,AR	99.97	518 of 524
SLC7A7	Lysinuric Protein Intolerance	AR	100	61 of 61
SPAG1	Ciliary Dyskinesia	AR	94.8	11 of 12
SPP1	Lupus Erythematosus	-	99.77	2 of 2

STAT4	Behçet Disease, Oligoarticular Idiopathic Arthritis, Lupus Erythematosus	-	99.98	4 of 4
STK36	Ciliary Dyskinesia	-	100	5 of 5
THBD	Hemolytic Uremic Syndrome, Thrombophilia	AD	99.91	29 of 30
ZMYND10	Ciliary Dyskinesia	AR	99.98	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 mutations according to HGMD

Methodology

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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