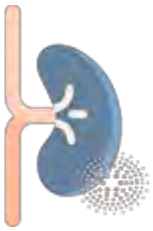
Nephrotic Syndrome

Precision Panel



Overview

Nephrotic Syndrome (NS) is defined as the presence of nephrotic-range proteinuria with a low serum albumin level, edema and hypercholesterolemia, indicating damage to the glomerular filtration barrier. Nephrotic-range proteinuria is known as the loss of 3.5 grams or more per day of protein in the urine. Nephrotic syndrome has a plethora of causes in which we can find primary kidney diseases such as minimal change disease, focal segmental glomerulosclerosis and membranous glomerulonephritis, among others. It can also be the result of systemic diseases that involve other organs apart from the kidney such as diabetes, lupus erythematosus and amyloidosis. Genetic causes of nephrotic syndrome consist of defects in glomerular filtration involving a variety of gene mutations, inherited in its majority in an autosomal recessive pattern.

The Igenomix Nephrotic Syndrome Precision Panel can be used to make a directed and accurate differential diagnosis of proteinuria 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 Nephrotic Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of Nephrotic Syndrome presenting with:

- Massive proteinuria (>3.5g/24 hours)
- Edema
- Thrombosis and embolic events
- Hypertension
- Increased susceptibility to infection
- Low albumin level
- Hyperlipidemia

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 treatment with a multidisciplinary team in the form of medical care with steroids, lipid lowering drugs, preventive anticoagulation or management of underlying systemic disease.
- Risk assessment of asymptomatic family members according to the mode of inheritance.
- Translation of genomic-informed medicine allowing for the improvement of the understanding of molecular anatomy of nephrotic syndrome and thus, the ability to care for patients.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ADA	Severe Combined Immunodeficiency, Omenn Syndrome	AR	100	97 of 98
ANLN	Focal Segmental Glomerulosclerosis	AD	99.45	6 of 6
APOA1	Familial Visceral Amyloidosis, Apolipoprotein A-1 Deficiency	AD	99.89	68 of 70
ARHGDIA	Nephrotic Syndrome	AR	99.41	3 of 3
ARL6	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 21
AVIL	Nephrotic Syndrome	AR	100	4 of 4
B2M	Familial Visceral Amyloidosis, Hypercatabolic Hypoproteinemia, Variant Abeta2m Amyloidosis	AD,AR	100	4 of 4
BBIP1	Bardet-Biedl Syndrome	AR	99.88	1 of 1
BBS1	Bardet-Biedl Syndrome	AR	100	102 of 105
BBS10	Bardet-Biedl Syndrome	AR	100	114 of 114
BBS12	Bardet-Biedl Syndrome	AR	99.78	61 of 61
BBS2	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	100	99 of 100
BBS4	Bardet-Biedl Syndrome	AR	100	45 of 48
BBS5	Bardet-Biedl Syndrome	AR	99.8	30 of 31
BBS7	Bardet-Biedl Syndrome	AR	100	48 of 48
BBS9	Bardet-Biedl Syndrome	AR	99.56	50 of 51
C1QBP	Combined Oxidative Phosphorylation Deficiency	AR	99.89	6 of 6
C3	Autosomal Recessive Complement Component 3 Deficiency, Atypical Hemolytic Uremic Syndrome	AD,AR	100	123 of 124
C8ORF37	Bardet-Biedl Syndrome, Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR,X,XR,G	na	na
CASP10	Autoimmune Lymphoproliferative Syndrome, Type IIa	AD	99.86	6 of 6
CCND1	Multiple Myeloma, Von Hippel-Lindau Syndrome	AD	99.95	1 of 1
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CHD7	Charge Syndrome, Omenn Syndrome	AD	96.25	823 of 896
CHST14	Musculocontractural Ehlers-Danlos Syndrome	AR	97.7	21 of 22
COL4A3	Autosomal Dominant Alport Syndrome, Autosomal Recessive Alport Syndrome, Benign Familial Hematuria	AD,AR	100	277 of 280
COL4A4	Autosomal Recessive Alport Syndrome, Benign Familial Hematuria	AD,AR	99.95	247 of 251
COL4A5	X-linked Alport Syndrome	X,XD,G	99.88	NA of NA
COQ2	Coenzyme Q10 Deficiency, Multiple System Atrophy, Leigh Syndrome With Nephrotic Syndrome	AD,AR	99.61	37 of 38
COQ6	Coenzyme Q10 Deficiency	AR	100	19 of 19
COQ8B	Nephrotic Syndrome	AR	100	NA of NA
CRB2	Ventriculomegaly With Cystic Kidney Disease, Focal Segmental Glomerulosclerosis	AR	99.5	26 of 29
CYBC1	Autosomal Recessive Chronic Granulomatous Disease	AR	na	na
DCLRE1C	Omenn Syndrome, Severe Combined Immunodeficiency With Sensitivity To Ionizing Radiation, Omenn Syndrome	AR	99.99	72 of 73
DGKE	Nephrotic Syndrome	AR	99.67	41 of 42
DHX37	46,XY Sex Reversal, 46,XY Partial Gonadal Dysgenesis, Neurodevelopmental Disorder With Brain Anomalies And With Or Without Vertebral Or Cardiac Anomalies, Testicular Regression Syndrome	AD,AR	99.87	13 of 13
DMRT3	46,XY Partial Gonadal Dysgenesis	-	88.67	1 of 1
EMP2	Familial Idiopathic Steroid-Resistant Nephrotic Syndrome	AR	99.98	3 of 3

FGA	Afibrinogenemia, Congenital Hypofibrinogenemia, Familial Visceral Amyloidosis, Familial Dysfibrinogenemia	AD,AR	100	153 of 154
FN1	Glomerulopathy With Fibronectin Deposits, Fibronectin Glomerulopathy	AD	100	34 of 34
FOXP3	Immunodysregulation, X-linked Polyendocrinopathy And Enteropathy	X,XR,G	99.86	NA of NA
GATA3	Hypoparathyroidism, Sensorineural Deafness, And Renal Disease	AD	100	81 of 81
GATA4	Testicular Anomalies With Or Without Congenital Heart Disease, 46,XY Partial Gonadal Dysgenesis, 8p23.1 Microdeletion Syndrome	AD	94.69	108 of 130
GLA	Fabry Disease	X,XR,G	98	NA of NA
GSN	Amyloidosis	AD	96.69	16 of 17
IFIH1	Aicardi-Goutieres Syndrome, Singleton-Merten Syndrome	AD	99.62	26 of 27
IFT172	Retinitis Pigmentosa, Bardet-Biedl Syndrome, Jeune Syndrome, Retinitis Pigmentosa	AR	100	37 of 37
IFT27	Bardet-Biedl Syndrome	AR	100	5 of 5
IL2RG	X-linked Severe Combined Immunodeficiency, Omenn Syndrome	X,XR,G	99.86	NA of NA
IL7R	Autosomal Recessive Severe Combined Immunodeficiency, Omenn Syndrome	AR	100	54 of 55
IRAK1	Pediatric Systemic Lupus Erythematosus	-	96.2	NA of NA
ITGA3	Interstitial Lung Disease, Nephrotic Syndrome, And Congenital Epidermolysis Bullosa	AR	99.2	11 of 11
JAK1	Autoinflammation, Immune Dysregulation, And Eosinophilia	AD	99.92	7 of 8
KANK2	Nephrotic Syndrome, Palmoplantar Keratoderma And Woolly Hair	AR	99.92	3 of 3
LAGE3	X-linked Galloway-Mowat Syndrome	X,XR,G	91.36	NA of NA
LAMB2	Nephrotic Syndrome With Or Without Ocular Abnormalities, Pierson Syndrome, Synaptic Congenital Myasthenic Syndromes	AR	100	129 of 129
LIG4	LIG4 Syndrome, Multiple Myeloma, Dubowitz Syndrome, Omenn Syndrome	AR	99.48	46 of 46
LMNB2	Barraquer-Simons Syndrome, Progressive Myoclonic Epilepsy, Acquired Partial Lipodystrophy	AD,AR	95.03	5 of 5
LMX1B	Nail-Patella Syndrome, 9q33.3q34.11 Microdeletion Syndrome, Nail-Patella-Like Renal Disease	AD	100	191 of 191
LYZ	Familial Visceral Amyloidosis	AD	100	10 of 10
LZTFL1	Bardet-Biedl Syndrome	AR	99.83	4 of 4
MAGI2	Nephrotic Syndrome	AR	93.82	7 of 9
MAP3K1	46,XY Sex Reversal, 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis	AD	96.5	31 of 32
MARS1	Charcot-Marie-Tooth Disease, Interstitial Lung And Liver Disease	AD,AR	99.98	19 of 19
MEFV	Familial Mediterranean Fever, Behçet Disease	AD,AR	96.77	174 of 188
MKKS	Bardet-Biedl Syndrome, Mckusick-Kaufman Syndrome	AR	89.96	71 of 71
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MME	Charcot-Marie-Tooth Disease, Spinocerebellar Ataxia, Congenital Membranous Nephropathy Due To Fetomaternal Anti-Neutral Endopeptidase Alloimmunization	AD,AR	100	33 of 33
MYO1E	Focal Segmental Glomerulosclerosis	AR	100	30 of 30
NLRP3	Cinca Syndrome, Familial Cold Inflammatory Syndrome, Keratoendotheliitis Fugax Hereditaria, Muckle-Wells Syndrome	AD	100	152 of 152
NPHP1	Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome, Joubert Syndrome With Renal Defect	AR	100	58 of 59
NPHS1	Congenital Nephrotic Syndrome	AR	100	351 of 352
NPHS2	Autosomal Recessive Steroid-Resistant Nephrotic Syndrome	AR	99.7	209 of 212
NR0B1	Congenital Adrenal Hypoplasia, 46,XX Testicular Disorder Of Sex Development, 46,XY Complete Gonadal Dysgenesis , 46,XY Partial Gonadal Dysgenesis	X,XR,G	99.87	NA of NA
NR5A1	46,XX Sex Reversal, 46,XY Sex Reversal, 46,XX Gonadal Dysgenesis, 46,XX Ovotesticular Disorder Of Sex Development, 46,XX Testicular Disorder Of Sex Development	AD	99.97	222 of 224
NUP107	Galloway-Mowat Syndrome, Nephrotic Syndrome, Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	99.91	15 of 15
NUP133	Galloway-Mowat Syndrome, Nephrotic Syndrome	AR	99.94	6 of 6
NUP160	Nephrotic Syndrome	AR	99.96	3 of 3
NUP205	Nephrotic Syndrome	AR	98.95	6 of 6
NUP85	Nephrotic Syndrome	AR	97.59	5 of 5
NUP93	Nephrotic Syndrome	AR	99.91	16 of 17
OSGEP	Galloway-Mowat Syndrome	AR	99.17	19 of 19
PAX2	Focal Segmental Glomerulosclerosis, Papillorenal Syndrome, Renal Coloboma Syndrome	AD	99.99	100 of 100
PDSS2	Primary Coenzyme Q10 Deficiency, Leigh Syndrome With Nephrotic Syndrome	AR	99.99	6 of 6
PLCE1	Nephrotic Syndrome	AR	99.93	73 of 73

PMM2	Congenital Disorder Of Glycosylation Type Ia	AR	100	127 of 129
PRKCD	Common Variable Immunodeficiency, Autoimmune Lymphoproliferative Syndrome	AR	100	9 of 9
PTPRO	Nephrotic Syndrome	AR	99.99	10 of 10
PUS3	Autosomal Recessive Mental Retardation, Severe Growth Deficiency-Strabismus-Extensive Dermal Melanocytosis-Intellectual Disability Syndrome	AR	99.01	8 of 9
RAG1	Combined Cellular And Humoral Immune Defects With Granulomas, Omenn Syndrome, Autosomal Recessive Severe Combined Immunodeficiency	AR	100	193 of 193
RAG2	Combined Cellular And Humoral Immune Defects With Granulomas, Omenn Syndrome, Autosomal Recessive Severe Combined Immunodeficiency	AR	100	90 of 91
RMRP	Anauxetic Dysplasia, Cartilage-Hair Hypoplasia, Metaphyseal Dysplasia Without Hypotrichosis, Omenn Syndrome	AR	na	na
SAA1	Amyloidosis	-	98.45	0 of 1
SCARB2	Action Myoclonus-Renal Failure Syndrome, Gaucher Disease Type 1, Unverricht-Lundborg Disease	AR	99.95	29 of 29
SDCCAG8	Bardet-Biedl Syndrome, Senior-Loken Syndrome	AR	96.29	18 of 19
SERPINA 1	Alpha-1-Antitrypsin Deficiency	AR	99.77	107 of 111
SGPL1	Nephrotic Syndrome	AR	98.96	18 of 18
SLC17A5	Infantile Sialic Acid Storage Disorder	AR	99.91	49 of 49
SLC35A2	X-linked Congenital Disorder Of Glycosylation	X,XD,G	99.97	NA of NA
SMARCA L1	Schimke Immuno-Osseous Dysplasia	AR	99.94	93 of 93
SNAP29	Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratodermasyndrome, Cednik Syndrome	AR	100	13 of 13
SOX9	Campomelic Dysplasia, 46,XX Ovotesticular Disorder Of Sex Development , 46,XX Testicular Disorder Of Sex Development , 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis, Isolated Pierre Robin Syndrome	AD	97.28	87 of 95
SPP1	Pediatric Systemic Lupus Erythematosus	-	99.77	2 of 2
SRY	46,XX Sex Reversal, 46,XY Sex Reversal, 45,X/46,XY Mixed Gonadal Dysgenesis, 46,XX Ovotesticular Disorder Of Sex Development , 46,XX Testicular Disorder Of Sex Development , 46,XY Complete Gonadal Dysgenesis , 46,XY Partial Gonadal Dysgenesis	X,XD,Y,G	45	NA of NA
STAT4	Behçet Disease, Pediatric Systemic Lupus Erythematosus		99.98	4 of 4
TBC1D8B	Nephrotic Syndrome	X,G	98.21	NA of NA
TBX18	Congenital Anomalies Of Kidney And Urinary Tract	AD	99.8	9 of 12
TP53RK	Galloway-Mowat Syndrome	AR	97.68	5 of 5
TPRKB	Galloway-Mowat Syndrome	AR	85.66	2 of 2
TRIM32	Bardet-Biedl Syndrome	AR	100	17 of 17
TRPC6	Focal Segmental Glomerulosclerosis	AD	99.92	52 of 55
TTC8	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	99.33	28 of 28
VAMP7	46,XY Partial Gonadal Dysgenesis		99.98	NA of NA
VPS33A	Mucopolysaccharidosis-Plus Syndrome	AR	97.86	1 of 1
WDPCP	Bardet-Biedl Syndrome, Heart Defect-Tongue Hamartoma-Polysyndactyly Syndrome, Meckel Syndrome	AR	99.3	8 of 8
WDR4	Galloway-Mowat Syndrome, Microcephaly, Growth Deficiency, Seizures, And Brain Malformations	AR	99.91	7 of 7
WDR73	Galloway-Mowat Syndrome, Camos Syndrome	AR	95.71	14 of 14
WT1	Denys-Drash Syndrome, Frasier Syndrome, Nephrotic Syndrome, Wilms Tumor, Aniridia, Genitourinary Anomalies, And Mental Retardation Syndrome Chromosome 11p13 Deletion Syndrome , 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis, Meacham Syndrome	AD	98.92	178 of 185
WWOX	46,XY Partial Gonadal Dysgenesis, Autosomal Recessive Cerebellar Ataxia-Epilepsy-Intellectual Disability Syndrome Due To Wwox Deficiency	AR	99.94	44 of 44
ZAP70	Multisystem Autoimmune Disease, Atypical Severe Combined Immunodeficiency	AR	99.99	30 of 30
ZFPM2	46,XY Sex Reversal, 46,XY Partial Gonadal Dysgenesis	AD	99.4	44 of 46
ZNF592	Camos Syndrome	-	99.93	1 of 1

*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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