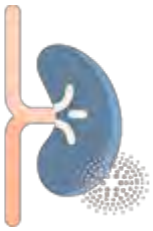
Polycystic Kidney Disease

Precision Panel



Overview

Polycystic Kidney Disease (PKD) is an inherited multisystemic and progressive disorder characterized by cyst formation and enlargement of the kidneys and other organs. Cysts are noncancerous round sacs containing fluid. These cysts eventually deteriorate renal anatomy and physiology causing them to lose function over time. Polycystic kidney disease is classified into two distinct disorders based on the inheritance pattern: autosomal dominant PKD (ADPKD) and autosomal recessive PKF (ARPKD). ARPKD is the most aggressive form and presents with severe pulmonary insufficiency and progressive renal failure with early onset during infancy. If left untreated, ARPKD is lethal before adolescence. ADPKD usually manifests during adulthood and is the most common inherited cause of chronic kidney disease. Cystic kidneys are common causes of end-stage renal disease, both in children and adults.

The Igenomix Polycystic Kidney Disease Precision Panel can be used to make a directed and accurate differential diagnosis of renal cysts 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 Polycystic Kidney Disease Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of polycystic kidneys presenting with:

- Gross hematuria
- Flank or abdominal pain
- Recurrent urinary tract infections
- Nephrolithiasis
- Palpable kidneys on abdominal exam
- Signs of chronic kidney disease (hypertension, fluid overload, uremia)
- Extrarenal cysts: hepatic, pancreatic, cerebral berry aneurysm
- Maternal oligohydramnios and Potter sequence

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 blood pressure control to prevent and delay end-stage renal disease, related complications and/or renal transplantation.
- Provide regular ultrasound and laboratory monitoring improving clinical management of patients, enhancing further with emerging therapeutic options.
- Genetic counselling session for risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ALG8	Polycystic Liver Disease With Or Without Kidney Cysts	AD,AR	99.5	22 of 22
ALG9	Polycystic Kidney Disease Potter Type I, With Microbrachycephaly, Hypertelorism, And Brachymelia	AR	99.99	6 of 6
ANKS6	Nephronophthisis	AR	93.45	17 of 17
ARVCF	22q11.2 Deletion Syndrome	-	99.95	2 of 2
BICC1	Cystic Renal Dysplasia, Autosomal Dominant Polycystic Kidney Disease	AD	99.89	5 of 5
CDC73	Parathyroid Carcinoma, Familial Isolated Hyperparathyroidism, Hyperparathyroidism-Jaw Tumor Syndrome	AD	100	95 of 95
COMT	22q11.2 Deletion Syndrome	AD	99.98	5 of 5
CPT2	Carnitine Palmitoyl Transferase II Deficiency	AD,AR	99.99	116 of 116
DNAJB11	Polycystic Kidney Disease With Or Without Polycystic Liver Disease	AD	99.89	6 of 6
DYNC2H1	Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome	AR,MU,D	99.78	214 of 221
DZIP1L	Autosomal Recessive Polycystic Kidney Disease	AR	99.83	5 of 5
ESCO2	Roberts Syndrome, Sc Phocomelia Syndrome	AR	99.69	32 of 32
ETFA	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	92.33	32 of 32
ETFB	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100	21 of 21
ETFDH	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100	221 of 222
EYA1	Branchiootorenal Syndrome, Otofaciocervical Syndrome, Bor Syndrome	AD	100	197 of 199
GANAB	Autosomal Dominant Polycystic Kidney Disease	AD	100	19 of 19
GATA3	Hypoparathyroidism, Sensorineural Deafness, And Renal Disease	AD	100	81 of 81
GLIS3	Neonatal Diabetes Mellitus With Congenital Hypothyroidism	AR	99.83	21 of 21
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Fetal And Neonatal Alloimmune Thrombocytopenia	AR	74.08	26 of 50
HIRA	22q11.2 Deletion Syndrome	-	99.99	5 of 5
IFT43	Cranioectodermal Dysplasia, Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia With Polydactyly	AR	100	6 of 6
JMJD1C	22q11.2 Deletion Syndrome	-	99.09	27 of 27
LRP5	Polycystic Liver Disease With Or Without Kidney Cysts, Van Buchem Disease Type 2, Isolated Polycystic Liver Disease	AD,AR	98.12	265 of 269
MKKS	Bardet-Biedl Syndrome, Mckusick-Kaufman Syndrome	AR	89.96	71 of 71
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome Type 1	AR	99.98	49 of 49
NEK1	Amyotrophic Lateral Sclerosis, Orofaciodigital Syndrome Type 2	AD,AR,MU,D	99.83	73 of 74
NPHP3	Meckel Syndrome, Nephronophthisis, Renal-Hepatic-Pancreatic Dysplasia, Senior-Loken Syndrome	AR	99.99	84 of 84
OFD1	Joubert Syndrome, Orofaciodigital Syndrome Type 1, Simpson-Golabi-Behmel Syndrome Type 2, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	NA of NA
PEX12	Peroxisome Biogenesis Disorder 3a (Zellweger), Infantile Refsum Disease, Zellweger Syndrome	AR	100	38 of 38

PEX5	Cerebrohepatorenal Syndrome, Infantile Refsum Disease, Zellweger Syndrome	AR	100	12 of 12
PKD1	Autosomal Dominant Polycystic Kidney Disease	AD	97.98	2078 of 2136
PKD2	Autosomal Dominant Polycystic Kidney Disease	AD	95.5	352 of 359
PKHD1	Autosomal Recessive Polycystic Kidney Disease	AR	99.97	582 of 585
RREB1	22q11.2 Deletion Syndrome	-	99.92	8 of 8
SEC24C	22q11.2 Deletion Syndrome	-	99.98	NA of NA
SHANK3	Phelan-Mcdermid Syndrome, Monosomy 22q13.3	AD,MU,P	96.67	NA of NA
SIX1	Branchiootorenal Syndrome, Autosomal Dominant Deafness, Bor Syndrome	AD	73	20 of 20
SKIV2L	Trichohepatoenteric Syndrome	AR	99.98	33 of 33
TBX1	DiGeorge Syndrome, Velocardiofacial Syndrome, 22q11.2 Deletion Syndrome, 22q11.2 Microduplication Syndrome	AD,AR	88.7	35 of 42
TMEM107	Meckel Syndrome, Orofaciodigital Syndrome XVI, Meckel Syndrome	AR	100	3 of 3
TMEM231	Joubert Syndrome With Oculorenal Defect, Meckel Syndrome, Orofaciodigital Syndrome Type 3	AR	98.63	20 of 21
TRIP11	Achondrogenesis Type Ia , Osteochondrodysplasia	AR	98.94	20 of 21
TSC1	Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	99.86	390 of 406
TSC2	Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	100	1157 of 1159
TTC37	Trichohepatoenteric Syndrome	AR	100	66 of 66
UFD1	22q11.2 Deletion Syndrome	-	99.98	NA of NA
WDR35	Cranioectodermal Dysplasia, Short-Rib Thoracic Dysplasia With Or Without Polydactyly	AR	100	31 of 33
ZNF423	Nephronophthisis, Joubert Syndrome With Oculorenal Defect	AD,AR	100	10 of 10

*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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