Oligohydramnios

Precision Panel

Overview

Oligohydramnios is defined as an abnormally low volume of amniotic fluid. Amniotic fluid is crucial for fetal development and growth, serving the foetus as protection from trauma and infection as well as helping in the development of fetal lungs. Normal amniotic fluid varies, on average it can reach up to 800-1000mL. An excess of amniotic fluid is termed polyhydramnios, decreased amniotic fluid is oligohydramnios. It occurs in approximately 11% of all pregnancies. Causes of oligohydramnios include rupture of membranes, fetal urinary tract blockage (renal agenesis, posterior urethral valves or polycystic kidney disease) which can have a genetic background associated with other genetic conditions. The mortality is high, especially if it is diagnosed during the first trimester as it can increase the risk for chest wall fixation and pulmonary hypoplasia.

The Igenomix Oligohydramnios Precision Panel can be used to make a directed and accurate differential diagnosis of oligohydramnios and uncover the genetics underlying this clinical sign ultimately leading to a better management and achieve a healthy baby at home. 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 Oligohydramnios Precision Panel is indicated for those patients with clinical and ultrasound findings of oligohydramnios presenting with:

- Abdominal discomfort
- Amniotic fluid index < 7cm
- Fetal structure survey revealing congenital abnormalities

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 for hospitalization and obstetric management in case of preterm delivery and provide adequate hydration.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

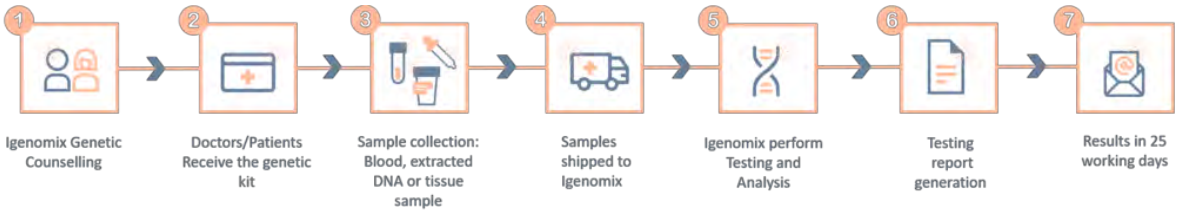
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE	HGMD**
ACE	Renal Tubular Dysgenesis	AR	88.42	40 of 44
AGT	Essential Hypertension, Renal Tubular Dysgenesis	AR,MU	100	16 of 18
AGTR1	Essential Hypertension, Renal Tubular Dysgenesis	AR,MU	100	8 of 8
ALB	Congenital Analbuminemia	AR	100	41 of 41
ALG8	Congenital Disorder Of Glycosylation Type Ih, Polycystic Liver Disease With Or Without Kidney Cysts	AD,AR	99.5	22 of 22
ALG9	Congenital Disorder Of Glycosylation, Type Il, Polycystic Kidney Disease Potter Type I, With Microbrachycephaly, Hypertelorism And Brachymelia	AR	99.99	6 of 6
ALX4	Craniosynostosis, Enlarged Parietal Foramina, Frontonasal Dysplasia-Alopecia-Genital Anomalies Syndrome, Isolated Scaphocephaly, Potocki-Shaffer Syndrome	AD,AR	99.94	25 of 25
AMER1	Osteopathia Striata With Cranial Sclerosis Syndrome	X,XD,G	99.45	NA of NA
ASCL1	Congenital Failure of Autonomic Control, Haddad Syndrome	AD	97.86	1 of 4
ATRX	Alpha-Thalassemia Myelodysplasia Syndrome, Mental Retardation-Hypotonic Facies Syndrome, Carpenter-Waziri Syndrome, Chudley-Lowry-Hoar Syndrome, Holmes-Gang Syndrome, Juberg-Marsidi Syndrome, Smith-Fineman-Myers Syndrome	X,XR,XD,G	98.5	NA of NA
B9D1	Joubert Syndrome, Meckel Syndrome	AR	90.23	11 of 11
B9D2	Meckel Syndrome	AR	84.81	4 of 5
BMPER	Diaphanospondylodysostosis	AR	99.98	22 of 22
BNC2	Congenital Lower Urinary Tract Obstruction, Posterior Urethral Valve	AD	98.85	14 of 14
BRCA1	Fanconi Anemia Complementation Group S	AD,AR,MU	98.97	2783 of 2894
BRCA2	Fanconi Anemia Complementation Group D1, Medulloblastoma, Wilms Tumor, Nephroblastoma	AD,AR,MU	98.51	3343 of 3451
BRIP1	Fanconi Anemia Complementation Group J	AD,AR	94.97	235 of 237
BUB1B	Mosaic Variegated Aneuploidy Syndrome	AD,AR	99.84	30 of 31
C1QBP	Combined Oxidative Phosphorylation Deficiency	AR	99.89	6 of 6
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome,	AR	99.43	98 of 100
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CEP55	Anhydramnios, Renal Dysplasia, Cerebellar Hypoplasia And Hydranencephaly, Meckel Syndrome	AR	99.22	3 of 3
CERT1	Autosomal Dominant Mental Retardation	AD	99.98	8 of 8
CHRM3	Absence of Abdominal Muscles With Urinary Tract Abnormality And Cryptorchidism, Prune Belly Syndrome	AR	99.8	4 of 4
CLTCL1			100	22 of 22
COG5	Congenital Disorder Of Glycosylation Type III	AR	100	19 of 19
COQ2	Coenzyme Q10 Deficiency , Multiple System Atrophy ,Leigh Syndrome With Nephrotic Syndrome	AD,AR	99.61	37 of 38
COQ7	Primary Coenzyme Q10 Deficiency	AR	99.71	6 of 6
CPT2	Carnitine Palmitoyltransferase II Deficiency	AD,AR	99.99	116 of 116
CSPP1	Joubert Syndrome, Meckel Syndrome	AR	98.32	29 of 30
DHPS	Neurodevelopmental Disorder With Seizures And Speech And Walking Impairment	AR	99.85	4 of 4
DOCK6	Adams-Oliver Syndrome	AR	98.06	37 of 37
DONSON	Microcephaly-Micromelia Syndrome	AR	98.14	26 of 27
EFEMP2	Autosomal Recessive Cutis Laxa	AR	99.99	17 of 17
ERCC4	Fanconi Anemia Complementation Group Q, Xeroderma Pigmentosum Complementation Group F, Progeroid Syndrome, Cockayne Syndrome Type 1	AR	99.68	69 of 72
ERGIC1	Arthrogryposis Multiplex Congenita	AR	100	2 of 2
EXOSC9	Pontocerebellar Hypoplasia Type 1d	AR	99.86	2 of 2
FANCA	Fanconi Anemia	AR	95.17	497 of 502
FANCB	Fanconi Anemia Complementation Group B, Vacterl Association With Hydrocephalus	X,XR,G	95.53	NA of NA
FANCC	Fanconi Anemia Complementation Group C	AR	100	75 of 75
FANCD2	Fanconi Anemia Complementation Group D2	AR	100	62 of 63
FANCE	Fanconi Anemia Complementation Group E	AR	97	17 of 18
FANCF	Fanconi Anemia Complementation Group F	AR	99.31	17 of 18
FANCG	Fanconi Anemia Complementation Group G		100	94 of 94
FANCI	Fanconi Anemia Complementation Group I	AR	100	53 of 54
FANCL	Fanconi Anemia Complementation Group L	AR	100	25 of 26
FANCM	Fanconi Anemia, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation	AR	99.73	59 of 61
FARSB	Rajab Interstitial Lung Disease With Brain Calcifications	AR	99.94	4 of 4
FBLN5	Autosomal Dominant and Recessive Cutis Laxa	AD,AR	97.43	23 of 23
FBN1	Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Lipodystrophy Syndrome, Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, Shprintzen-Goldberg Syndrome	AD	100	2836 of 2845
FGF20	Bilateral Renal Hypodysplasia/Aplasia	AR	99.76	2 of 2
GATA6	Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of Fallot	AD,AR	84.19	66 of 84
GLI3	Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome	AD,AR	100	231 of 231
GMPPB	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Myasthenic Syndromes With Glycosylation Defect, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease	AR	99.95	53 of 53
GNPTAB	Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta	AR	100	279 of 280
GREB1L	Bilateral Renal Hypodysplasia/Aplasia	AD	97.94	41 of 41

HBA1	Alpha-thalassemia, Heinz Body Anemia , Hemoglobin H Disease, Hb Bart's Hydrops Fetalis	AD	98.87	125 of 152
HBA2	Alpha-thalassemia, Heinz Body Anemias, Hemoglobin H Disease, Hb Bart's Hydrops Fetalis	AD	74.46	118 of 231
HNF1B	Noninsulin-Dependent Diabetes Mellitus, Renal Cysts And Diabetes Syndrome, Hnf1b-Related Autosomal Dominant Tubulointerstitial Kidney Disease	AD	100	219 of 220
HSPA9	Sideroblastic Anemia, Even-Plus Syndrome	AD,AR	99.72	14 of 14
HYMAI	Transient Neonatal Diabetes Mellitus, Paternal Uniparental Disomy Of Chromosome	AD	na	na
INVS	Nephronophthisis, Senior-Loken Syndrome	AR	99.9	38 of 38
ITGA8	Bilateral Renal Hypodysplasia/Aplasia	AR	99.68	7 of 7
KIF14	Meckel Syndrome, Autosomal Recessive Primary Microcephaly	AR	99.84	18 of 18
LARS2	Hydrops, Lactic Acidosis, And Sideroblastic Anemia, Perrault Syndrome	AR	99.99	20 of 20
LHX1	17q12 Microdeletion Syndrome		100	6 of 6
LIFR	Stuve-Wiedemann Syndrome	AR	99.81	33 of 33
MAD2L2	Fanconi Anemia Complementation Group V	AR	99.91	1 of 1
MBTPS2	Ichthyosis Follicularis, Atrichia, And Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, Osteogenesis Imperfecta, Type XIX, Bresek Syndrome	X,XR,G	100	NA of NA
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MYH3	Arthrogryposis, Contractures, Pterygia And Spondylocarpostarsal Fusion Syndrome, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome , Sheldon-Hall Syndrome	AD,AR	100	46 of 47
NALCN	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental Delay, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome	AD,AR	99.97	69 of 69
NEK8	Nephronophthisi, Renal-Hepatic-Pancreatic Dysplasia	AR	100	24 of 24
NEK9	Arthrogryposis, Perthes Disease, And Upward Gaze Palsy, Lethal Congenital Contracture Syndrome	AR	99.98	4 of 4
NPHP3	Meckel Syndrome, Nephronophthisis, Renal-Hepatic-Pancreatic Dysplasia , NPHP3-Related Meckel-like Syndrome, Senior-Loken Syndrome	AR	99.99	84 of 84
OSGEP	Galloway-Mowat Syndrome	AR	99.17	19 of 19
PALB2	Fanconi Anemia Complementation Group N	AD,AR	98.78	601 of 617
PBX1	Congenital Anomalies Of Kidney And Urinary Tract Syndrome With Or Without Hearing Loss, Abnormal Ears, Or Developmental Delay	AD	98	18 of 18
PDSS2	Coenzyme Q10 Deficiency, Leigh Syndrome With Nephrotic Syndrome	AR	99.99	6 of 6
PGAP2	Hyperphosphatasia With Mental Retardation Syndrome	AR	99.99	11 of 11
PGAP3	Hyperphosphatasia With Mental Retardation Syndrome	AR	97	19 of 20
PHOX2B	Congenital Failure of Autonomic Control, Neuroblastoma With Hirschsprung Disease, Haddad Syndrome, Ondine Syndrome	AD	90.74	58 of 71
PIGL	Zunich Neuroectodermal Syndrome, Chime Syndrome, Hyperphosphatasia-Intellectual Disability Syndrome	AR	86	11 of 13
PIGO	Hyperphosphatasia With Mental Retardation Syndrome	AR	99.93	21 of 21
PIGV	Hyperphosphatasia With Mental Retardation	AR	99.99	16 of 16
PIGW	Hyperphosphatasia With Mental Retardation Syndrome	AR	99.52	6 of 6
PIGY	Hyperphosphatasia With Mental Retardation Syndrome	AR	100	1 of 2
PKHD1	Autosomal Recessive Polycystic Kidney Disease	AR	99.97	582 of 585
PLAGL1	Paternal Uniparental Disomy Of Chromosome		95.56	2 of 2
POR	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency	AD,AR	99.98	67 of 68
PUF60	Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome	AD	100	30 of 30
RAD51	Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements	AD	99.98	16 of 16
RAD51C	Fanconi Anemia Complementation Group O	AR	100	130 of 130
REN	Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis	AD,AR	100	23 of 23
RET	Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis	AD	100	453 of 454
RFWD3	Fanconi Anemia Complementation Group W	AR	99.99	2 of 2
RNU4ATAC	Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome	AR	na	na
RPGRIP1	Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome	AR	99.33	146 of 159
RPGRIP1L	Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome	AR	99.96	52 of 52
SEC24D	Cole-Carpenter Syndrome	AR	99.97	14 of 14
SLC25A24	Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome	AD	99.59	2 of 2
SLX4	Fanconi Anemia Complementation Group P	AR	99.92	76 of 76
TALDO1	Transaldolase Deficiency	AR	95	13 of 14
TBCK	Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies	AR	99.95	15 of 15
TCTN2	Joubert Syndrome, Meckel Syndrome	AR	100	14 of 14
TCTN3	Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI	AR	99.99	13 of 13
TMEM107	Meckel Syndrome, Orofaciodigital Syndrome XVI, Meckel Syndrome	AR	100	3 of 3
TMEM216	Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI	AR	98.74	8 of 8
TMEM231	Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type III	AR	98.63	20 of 21
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome	AR	96.93	177 of 179
TMEM70	Mitochondrial Complex V (ATP Synthase) Deficiency, TMEM70-Related Mitochondrial Encephalo-Cardio-Myopathy	AR	100	22 of 24
TRIP4	Congenital Muscular Dystrophy, Spinal Muscular Atrophy With Congenital Bone Abnormalities	AR	99.92	3 of 3
UBE2A	X-linked Syndromic Mental Retardation	X,XR,G	99.99	NA of NA
UBE2T	Fanconi Anemia Complementation Group T	AR	100	4 of 4
WDPCP	Bardet-Biedl Syndrome, Congenital Heart Defects, Hamartomas Of Tongue, And Polysyndactyly, Meckel Syndrome	AR	99.3	8 of 8
WDR73	Galloway-Mowat Syndrome, Camos Syndrome	AR	95.71	14 of 14

WNT4	46,XX Sex Reversal With Dysgenesis Of Kidneys, Adrenals, And Lungs, Mullerian Aplasia And Hyperandrogenism, Serkal Syndrome	AD,AR	100	8 of 8
XRCC2	Fanconi Anemia Complementation Group U	AR	98.39	28 of 28

*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



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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.

References

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