



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

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FANCF FANCG FANCOI Anemia Complementation Group F FANCO FANCOI Fanconi Anemia Complementation Group G 100 94 of 94					
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FANCM Fanconi Anemia, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation FARSB Rajab Interstitial Lung Disease With Brain Calcifications ALOSOMAI Dominant and Recessive Cutis Laxa AUGOSMAI Dominant and Recessive Cutis Laxa ACOMICIC Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Lipodystrophy Syndrome, Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, FBN1 Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, Shprintzen-Goldberg Syndrome Bilateral Renal Hypodysplasia/Aplasia Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of Fallot Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome 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 GNPTAB Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta AR 99.76 2 of 2 2 of 2 2 of 2 2 of 2 3 of 23 4 AR 99.76 2 of 2 4 AR 99.76 2					53 of 54
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FBLN5Autosomal Dominant and Recessive Cutis LaxaAD,AR97.4323 of 23Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Lipodystrophy Syndrome, Marfan Syndrome, Marfan Syndrome, Stifff Skin Syndrome, Weill-Marchesani Syndrome, Shprintzen-Goldberg SyndromeAD1002836 of 2845FGF20Bilateral Renal Hypodysplasia/AplasiaAR99.762 of 2Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of FallotAD,AR84.1966 of 84GLI3Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Myasthenic Syndromes With AR4D,AR99.9553 of 53GNPTABMucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/BetaAR100279 of 280	FANCM		AR	99.73	59 of 61
Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Lipodystrophy Syndrome, Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, Shprintzen-Goldberg Syndrome Bilateral Renal Hypodysplasia/Aplasia Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of Fallot Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome 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 GNPTAB Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta AR 100 2836 of 2845 2845 2845 2846 2847 2845 2845 2845 2845 2845 2845 2845 2845	FARSB	Rajab Interstitial Lung Disease With Brain Calcifications	AR	99.94	4 of 4
FBN1Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Shprintzen-Goldberg SyndromeAD1002835 of 2845FGF20Bilateral Renal Hypodysplasia/AplasiaAR99.762 of 2GATA6Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of FallotAD,AR84.1966 of 84GLI3Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal SyndromeAD,AR100231 of 231 of 231GMPPBMuscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Congenital Myasthenic Syndromes With Glycosylation Defect, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain DiseaseAR99.9553 of 53 of 53GNPTABMucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/BetaAR100279 of 280	FBLN5	Autosomal Dominant and Recessive Cutis Laxa	AD,AR	97.43	23 of 23
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 Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome 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 GNPTAB Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta AR 99.76 2 of 2 AD,AR 84.19 66 of 84 AD,AR 100 231 of 23	FBN1	Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome,	AD	100	
Atrial Septal Defect, Atrioventricular Septal Defect, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of Fallot Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome 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 GNPTAB Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta AR 100 279 of 280	ECESO		A D	00.70	2 04 2
Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of Fallot Greig Cephalopolysyndactyly Syndrome, Congenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome 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 GNPTAB Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta AR 100 279 of 280		* * * * * * * * * * * * * * * * * * * *			
Pallister-Hall Syndrome, Acrocallosal Syndrome Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Myasthenic Syndromes With AR 99.95 53 of 53 Glycosylation Defect, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta AR 100 279 of 280	GATA6	Arteriosus Communis, Pancreatic Agenesis And Congenital Heart Defects, Tetralogy Of Fallot	AD,AR	84.19	
GMPPBMuscular Dystrophy With Cerebellar Involvement, Congenital Myasthenic Syndromes With Glycosylation Defect, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain DiseaseAR99.9553 of 53GNPTABMucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/BetaAR100279 of 280	GLI3	Pallister-Hall Syndrome, Acrocallosal Syndrome	AD,AR	100	
Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta Ak 100 280	GMPPB	Muscular Dystrophy With Cerebellar Involvement, Congenital Myasthenic Syndromes With	AR	99.95	53 of 53
	GNPTAB	Mucolipidosis II Alpha/Beta, Mucolipidosis III Alpha/Beta	AR	100	
	GREB1L	Bilateral Renal Hypodysplasia/Aplasia	AD	97.94	





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HBA1	Alpha-thalassemia, Heinz Body Anemia , Hemoglobin H Disease, Hb Bart's Hydrops Fetalis	AD	98.87	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	AD	100	219 of
HSPA9	Autosomal Dominant Tubulointerstitial Kidney Disease		99.72	220 14 of 14
HYMAI	Sideroblastic Anemia, Even-Plus Syndrome Transient Neonatal Diabetes Mellitus, Paternal Uniparental Disomy Of Chromosome	AD,AR AD	99.72 na	14 01 14 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 LHX1	Hydrops, Lactic Acidosis, And Sideroblastic Anemia, Perrault Syndrome 17q12 Microdeletion Syndrome	AR	99.99 100	20 of 20 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	X,XR,G	100	NA of NA
MKS1	Decalvans, Osteogenesis Imperfecta, Type XIX, Bresek Syndrome Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
	Arthrogryposis, Contractures, Pterygia And Spondylocarpostarsal Fusion Syndrome, Digitotalar			
МҮН3	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,	AD,AR	99.97	69 of 69
NEK8	Digitotalar Dysmorphism, Freeman-Sheldon Syndrome Nephronophthisi, Renal-Hepatic-Pancreatic Dysplasia	AR	100	24 of 24
	Arthrogryposis, Perthes Disease, And Upward Gaze Palsy, Lethal Congenital Contracture			
NEK9	Syndrome	AR	99.98	4 of 4
NPHP3	Meckel Syndrome, Nephronophthisis, Renal-Hepatic-Pancreatic Dysplasia , NPHP3-Related	AR	99.99	84 of 84
OSGEP	Meckel-like Syndrome, Senior-Loken Syndrome Galloway-Mowat Syndrome	AR	99.17	19 of 19
				601 of
PALB2	Fanconi Anemia Complementation Group N	AD,AR	98.78	617
PBX1	Congenital Anomalies Of Kidney And Urinary Tract Syndrome With Or Without Hearing Loss,	AD	98	18 of 18
PDSS2	Abnormal Ears, Or Developmental Delay 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	AD	90.74	58 of 71
	Syndrome, Ondine Syndrome Zunich Neuroectodermal Syndrome, Chime Syndrome, Hyperphosphatasia-Intellectual Disability			
PIGL	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 PIGY	Hyperphosphatasia With Mental Retardation Syndrome	AR AR	99.52 100	6 of 6 1 of 2
	Hyperphosphatasia With Mental Retardation Syndrome			582 of
PKHD1	Autosomal Recessive Polycystic Kidney Disease	AR	99.97	585
DIACIA				
PLAGL1	Paternal Uniparental Disomy Of Chromosome		95.56	2 of 2
PLAGLT	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital	AD,AR	95.56 99.98	
POR	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency		99.98	2 of 2 67 of 68
POR PUF60	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital	AD	99.98 100	2 of 2
POR	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-		99.98	2 of 2 67 of 68 30 of 30 16 of 16
POR PUF60	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome	AD	99.98 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of
POR PUF60 RAD51	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements	AD AD	99.98 100 99.98	2 of 2 67 of 68 30 of 30 16 of 16
POR PUF60 RAD51 RAD51C REN	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O	AD AD AR AD,AR	99.98 100 99.98 100 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130
POR PUF60 RAD51 RAD51C REN RET	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis	AD AD AR AD,AR AD	99.98 100 99.98 100 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454
POR PUF60 RAD51 RAD51C REN RET RFWD3	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W	AD AD AR AD,AR AD	99.98 100 99.98 100 100 100 99.99	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2
POR PUF60 RAD51 RAD51C REN RET	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis	AD AD AR AD,AR AD	99.98 100 99.98 100 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome	AD AR AD,AR AD AR AD	99.98 100 99.98 100 100 100 99.99 na	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome	AD AR AD,AR AD AR AD AR AR AR	99.98 100 99.98 100 100 100 99.99 na	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome	AD AR AD,AR AD AR AR AR AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome	AD AR AD,AR AD AR AR AR AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97	2 of 2 67 of 68 30 of 30 16 of 16 130 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D SLC25A24	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome	AD AR AD,AR AD AR AR AR AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97	2 of 2 67 of 68 30 of 30 16 of 16 130 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC25A24 SLX4	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 SEC24D SLC25A24 SLX4 TALD01	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coae-Carpenter Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency	AD AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D SLC25A24 SLX4 TALD01 TBCK	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies	AD AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 99.95	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 of 130 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1L SEC24D SLC25A24 SLX4 TALDO1 TBCK TCTN2	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Meckel Syndrome	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 90.95 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D SLC25A24 SLX4 TALD01 TBCK TCTN2 TCTN3	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Meckel Syndrome Joubert Syndrome, Meckel Syndrome Joubert Syndrome, Meckel Syndrome Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 99.95 100 99.99	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 14
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 SEC24D SLC25A24 SLX4 TALD01 TBCK TCTN2 TCTN3 TMEM107	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Meckel Syndrome Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type III	AD AD AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 100 99.99 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 SEC24D SLC25A24 SLX4 TALD01 TBCK TCTN2 TCTN3 TMEM107 TMEM216	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome XVI, Meckel Syndrome Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Ocoach Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Ocoach Syndrome, Joubert Syndrome, Meckel Syndrome, Meckel Syndrome, Ocoach Syndrome, Joubert Syndrome, Meckel Syndrome, Meckel Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert S	AD AD AR AD AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 99.95 100 99.99 100 98.74	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1L SEC24D SLC25A24 SLX4 TALDO1 TBCK TCTN2 TCTN3 TMEM107 TMEM216 TMEM231 TMEM67	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Meckel Syndrome Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type III Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Neckel Syndrome, Nephronophthisis, Rhyns Syndrome	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 100 99.99 100 98.74 98.63 96.93	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 SEC24D SLC25A24 SLX4 TALD01 TBCK TCTN2 TCTN3 TMEM107 TMEM216 TMEM231	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome XVI, Meckel Syndrome Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Ocoach Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Ocoach Syndrome, Joubert Syndrome, Meckel Syndrome, Meckel Syndrome, Ocoach Syndrome, Joubert Syndrome, Meckel Syndrome, Meckel Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert Syndrome, Joubert S	AD AD AR AD AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 90.95 100 98.74 98.63	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1L SEC24D SLC25A24 SLX4 TALDO1 TBCK TCTN2 TCTN3 TMEM107 TMEM216 TMEM231 TMEM67	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type III Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Neokel Syndrome, Nephronophthisis, Rhyns Syndrome Mitochondrial Complex V (ATP Synthase) Deficiency, TMEM70-Related Mitochondrial	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 100 99.99 100 98.74 98.63 96.93	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 SEC24D SLC25A24 SLX4 TALDO1 TBCK TCTN2 TCTN3 TMEM107 TMEM216 TMEM67 TMEM70	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome XVI, Meckel Syndrome Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type III Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome Mitochondrial Complex V (ATP Synthase) Deficiency, TMEM70-Related Mitochondrial Encephalo-Cardio-Myopathy	AD AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 99.95 100 99.99 100 98.74 98.63 96.93 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of 179 22 of 24
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POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D SLC25A24 SLX4 TALDO1 TALDO1 TCTN3 TMEM107 TMEM216 TMEM231 TMEM67 TMEM70 TRIP4 UBE2A UBE2T	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Coach Syndrome, Meckel Syndrome, Joubert Syndrome With Renal Defect, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome XVI, Meckel Syndrome Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome, Doubert Syndrome, Neckel Syndrome, N	AD AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 90.92 95 90.95 100 99.99 100 98.74 98.63 96.93 100 99.92 99.99 100	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of 179 22 of 24 3 of 3 NA of NA 4 of 4
POR PUF60 RAD51 RAD51C REN RET RFWD3 RNU4ATAC RPGRIP1 RPGRIP1L SEC24D SLC25A24 SLX4 TALD01 TBCK TCTN2 TCTN3 TMEM107 TMEM216 TMEM67 TMEM70 TRIP4 UBE2A	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability-Cardiac Anomalies-Short Stature-Joint Laxity Syndrome Fanconi Anemia Complementation Group R, Familial Congenital Mirror Movements Fanconi Anemia Complementation Group O Familial Juvenile Hyperuricemic Nephropathy, Renal Tubular Dysgenesis Congenital Failure of Autonomic Control, Hirschsprung Disease, Haddad Syndrome, Hirschsprung Disease, Bilateral Renal Agenesis Fanconi Anemia Complementation Group W Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome Cole-Carpenter Syndrome Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome Fanconi Anemia Complementation Group P Transaldolase Deficiency Infantile Hypotonia With Psychomotor Retardation And Characteristic Facies Joubert Syndrome, Meckel Syndrome Joubert Syndrome, Orofaciodigital Syndrome IV, Orofaciodigital Syndrome Type VI Meckel Syndrome, Orofaciodigital Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome With Oculorenal Defect, Orofaciodigital Syndrome Type VI Meckel Syndrome, Joubert Syndrome, Joubert Syndrome, Meckel Syndrome, Neckel Syndrome, Nephronophthisis, Rhyns Syndrome Mitochondrial Complex V (ATP Synthase) Deficiency, TMEM70-Related Mitochondrial Encephalo-Cardio-Myopathy Congenital Muscular Dystrophy, Spinal Muscular Atrophy With Congenital Bone Abnormalities X-linked Syndromic Mental Retardation Fanconi Anemia Complementation G	AD AR AD,AR AD AR	99.98 100 99.98 100 100 100 100 99.99 na 99.33 99.96 99.97 99.59 99.92 95 100 99.99 100 98.74 98.63 96.93 100 99.92 99.99	2 of 2 67 of 68 30 of 30 16 of 16 130 of 130 23 of 23 453 of 454 2 of 2 na 146 of 159 52 of 52 14 of 14 2 of 2 76 of 76 13 of 14 15 of 15 14 of 14 13 of 13 3 of 3 8 of 8 20 of 21 177 of 179 22 of 24 3 of 3 NA of NA





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.

Methodology





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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^{**}Number of clinically relevant mutations according to HGMD