Infertility

Precision Panel

Overview

Infertility is defined as the failure to conceive, regardless of the cause, after 1 year of unprotected intercourse. This condition affects approximately 10-15% of reproductive-aged couples. Infertility can be caused by a female factor, male factor or environmental. The female etiology of infertility can be cervical, uterine, ovarian, tubal or peritoneal. The male factors that affect fertility include pretesticular, testicular or post-testicular. Environmental factors that affect fertility include excessive exercise, occupational, toxic substances, inadequate diet associated with extreme weight loss or gain as well as advanced age. In our current society, some women postpone childbearing until their 30s and beyond, so they tend to have more difficulty conceiving and increased risk of miscarriage. Clinically, it is a highly heterogeneous pathology with a complex etiology that includes environmental and genetic factors. It is estimated that nearly 50% of infertility cases are due to genetic defects.

The Igenomix Infertility Precision Panel can be used to make a directed and accurate differential diagnosis of inability to conceive 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 Infertility Precision Panel is indicated for those patients with clinical suspicion of infertility presenting with the following manifestations:

- Inability to conceive after 1 year of unprotected intercourse
- Family history of infertility
- Recurrent miscarriages
- Family history of recurrent miscarriages

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 an initial consultation and workup, early pharmacologic and surgical treatment and assisted reproductive technologies (ART).
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ADGRG2	Congenital Bilateral Absence Of Vas Deferens	X,G	99.64%	NA of NA
AMH	Persistent Mullerian Duct Syndrome, Types I And II	AR	98.17%	76 of 96
AMHR2	Persistent Mullerian Duct Syndrome, Types I And II	AR	100%	87 of 95
ANOS1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	X,XR,G	96.86%	NA of NA
AR	Androgen Insensitivity Syndrome, X-linked hypospadias, Kennedy Disease, Partial Androgen Insensitivity Syndrome, Prostate Cancer, Reifenstein Syndrome, X-linked Spinal And Bulbar Muscular Atrophy	AD,X,XR,G	97.96%	NA of NA
ARL13B	Joubert Syndrome	AR	99.77%	10 of 10
ARL6	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AD,AR,X,XR,G	100%	17 of 21
AURKC	Male Infertility With Large-Headed, Multiflagellar, Polyploid Spermatozoa	AR	100%	5 of 5
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	99.80%	30 of 31
BBS7	Bardet-Biedl Syndrome	AR	100%	48 of 48
BBS9	Bardet-Biedl Syndrome	AR	99.56%	50 of 51
BMP15	46,XX Gonadal Dysgenesis, Ovarian Dysgenesis	X,G	98.05%	NA of NA
C8ORF37	Bardet-Biedl Syndrome, Cone Rod Dystrophy, Retinitis Pigmentosa	AD,AR,X,XR,G	na	na
CAPN10	Noninsulin-Dependent Diabetes Mellitus, Metabolic Syndrome, Polycystic Ovarian Syndrome	-	99.92%	4 of 4
CATSPER1	Spermatogenic Failure	AR	99.97%	4 of 4
CATSPER2	Deafness-Infertility Syndrome	AR	99.87%	1 of 1
CBX2	46,XY Complete Gonadal Dysgenesis, 46XY Sex Reversal	AR	100%	6 of 6
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43%	98 of 100
CCDC28B	Bardet-Biedl Syndrome	AR	99.83%	1 of 1
CEP164	Nephronophthisis, Senior-Loken Syndrome	AR	99.98%	10 of 10
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47%	293 of 327
CFTR	Bronchiectasis, Congenital Bilateral Absence Of Vas Deferens, Cystic Fibrosis, Hereditary Chronic Pancreatitis, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation	AD,AR	95.45%	1615 of 1730
CHD7	CHARGE Syndrome, Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallman Syndrome, Omenn Syndrome	AD	96.25%	823 of 896
CYP11A1	46,XY Disorder Of Sex Development-Adrenal Insufficiency Due To Cyp11A1 Deficiency, Congenital Adrenal Insufficiency With 46,XY Sex Reversal	-	100%	39 of 39
CYP17A1	46,XY Disorder Of Sex Development Due To Isolated 17,20-Lyase Deficiency, Congenital Adrenal Hyperplasia Due To 17-Alpha-Hydroxylase Deficiency	AR	100%	127 of 127
CYP19A1	Aromatase Deficiency, Aromatase Excess Syndrome	AD,AR	100%	33 of 35
CYP21A2	Congenital Adrenal Hyperplasia Due To 21-Hydroxylase Deficiency	AR	99.98%	278 of 280
DENND1A	Polycystic Ovarian Syndrome	-	98.61%	NA of NA
DHH	46XY Complete Gonadal Dysgenesis, 46XY Gonadal Dysgenesis-Motor And Sensory Neuropathy Syndrome, 46XY Sex Reversal	AR	99.85%	21 of 21
DIAPH2	Premature Ovarian Failure	X,XD,G	98.66%	NA of NA
DMC1	Infertility Due to Meiotic and/or Mitotic Failure	-	100%	2 of 2
DNAH1	Primary Ciliary Dyskinesia, Spermatogenic Failure	AR	100%	58 of 58
DNAH5	Primary Ciliary Dyskinesia With Or Without Situs Inversus, Primary Ciliary Dyskinesia	AR	100%	277 of 278
DNAI1	Kartagener Syndrome, Primary Ciliary Dyskinesia	AR	96.91%	43 of 43
DPY19L2	Spermatogenic Failure	AR	97.65%	16 of 20
DUSP6	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.36%	4 of 4
ENPP1	Generalized Arterial Calcification Of Infancy, Autosomal Recessive Hypophosphatemic Rickets, Cole Disease, Noninsulin-dependent Diabetes Mellitus, Autosomal Recessive Hypophosphatemic Rickets, Obesity, Pseudoxanthoma Elasticum	AD,AR,MU,P	96.59%	73 of 75
ESR1	Breast Cancer, Estrogen Resistance Syndrome, Migraine With Or Without Aura	AD,AR	99.98%	14 of 14

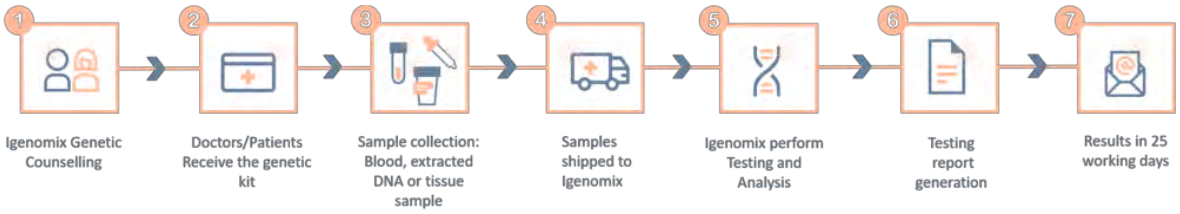
F2	Congenital Factor II Deficiency, Congenital Prothrombin Deficiency, Ischemic Stroke, Pregnancy Loss, Venous Thromboembolism	AD,AR,MU	100%	66 of 66
F5	Budd-Chiari Syndrome, Memembranous Obstruction Of Inferior Vena Cava, Congenital Factor V Deficiency, Ischemic Stroke, Recurrent Pregnancy Loss, Thrombophilia Due To Deficiency Of Activated Protein C Cofactor	AD,AR,MU	99.99%	165 of 167
FEZF1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AR	99.95%	3 of 3
FGF17	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.98%	8 of 8
FGF8	Alobar Holoprosencephaly, Kallmann Syndrome, Lobar Holoprosencephaly, Microform Holoprosencephaly, Midline Interhemispheric Variant Of Holoprosencephaly, Normosmic Congenital Hypogonadotropic Hypogonadism, Septopreoptic Holoprosencephaly	AD	98.36%	38 of 38
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Isolated Trigonocephaly, Jackson-Weiss Syndrome, Kallmann Syndrome, Lobar Holoprosencephaly, Microform Holoprosencephaly, Normosmic Congenital Hypogonadotropic Hypogonadism, Oligodontia, Osteoglophonic Dysplasia, Pfeiffer Syndrome, Septo-Optic Dysplasia Spectrum, Nonsyndromic Trigonocephaly	AD	100%	279 of 280
FIGLA	Premature Ovarian Failure	AD	98.47%	4 of 5
FLRT3	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallman Syndrome	AD	99.98%	7 of 7
FMR1	Fragile X Mental Retardation Syndrome, Fragile X-Associated Tremor/Ataxia Syndrome, Premature Ovarian Failure, XQ27.3q28 Duplication Syndrome	X,XD,G	99.80%	NA of NA
FOXL2	Blepharophimosis, Ptosis And Epicanthus Inversus, Premature Ovarian Failure	AD	89.36%	136 of 201
FOXO3	Chromosome 6Q Deletion, Rhabdomyosarcoma	-	95.67%	NA of NA
FSHB	Hypogonadotropic Hypogonadism Without Anosmia, Isolated Follicle Stimulating Hormone Deficiency	AR	100%	8 of 8
FSHR	46,XX Gonadal Dysgenesis, Ovarian Dysgenesis, Ovarian Hyperstimulation Syndrome	AD,AR	100%	41 of 43
GALNTL5	Male Infertility With Impairment of Sperm Motility		99.95%	2 of 2
GALT	Classic Galactosemia	AR	100%	350 of 350
GDF9	Premature Ovarian Failure	AR	100%	13 of 13
GNRH1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Normosmic Congenital Hypogonadotropic Hypogonadism	AR	100%	12 of 12
GNRHR	Hypogonadotropic Hypogonadism Without Anosmia, Normosmic Congenital Hypogonadotropic Hypogonadism	AR	100%	59 of 59
HESX1	Combined Pituitary Hormone Deficiencies, Hypothyroidism, Kallmann Syndrome, Pituitary Stalk Interruption Syndrome, Septo-Optic Dysplasia Spectrum	AD,AR	100%	26 of 26
HEXA	Tay-Sachs Disease	AR	100%	205 of 206
HFE	Alzheimer Disease, Hemochromatosis, Porphyria Cutanea Tarda, Porphyria Variegata	AD,AR	100%	55 of 57
HFM1	Premature Ovarian Failure	AR	99.17%	10 of 10
HS6ST1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD	99.97%	8 of 8
HSD17B3	46,XY Disorder Of Sex Development Due To 17-Beta-Hydroxysteroid Dehydrogenase 3 Deficiency, Male Pseudohermaphroditism	AR	100%	61 of 61
HSF2	Familial Erythroleukemia, Y-linked Spermatogenic Failure		99.90%	1 of 1
IFT172	Bardet-Biedl Syndrome, Jeune Syndrome, Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia With Or Without Polydactyly	AR	100%	37 of 37
IFT27	Bardet-Biedl Syndrome	AR	100%	5 of 5
IL17RD	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD,AR	99.95%	17 of 17
INPP5E	Joubert Syndrome, Mental Retardation, Truncal Obesity, Retinal Dystrophy, And Micropenis	AR	99.89%	56 of 56
KIF7	Acrocallosal Syndrome, Hydrolethalus Syndrome, Macrocephaly With Multiple Epiphyseal Dysplasia And Distinctive Facies, Orofaciodigital Syndrome	AR	94.91%	47 of 50
KISS1	Hypogonadotropic Hypogonadism With Or Without Anosmia, Normosmic Congenital Hypogonadotropic Hypogonadism	AR	100%	9 of 10
KISS1R	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism, Central Precocious Puberty	AD,AR	99.41%	42 of 43
KLHL10	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AD	99.98%	5 of 5
LEP	Obesity Due To Congenital Leptin Deficiency	AR	100%	19 of 19
LEPR	Obesity Due To Leptin Receptor Gene Deficiency	AR	97.92%	49 of 49
LHB	Hypogonadotropic Hypogonadism Without Anosmia	AR	100%	11 of 11
LHCGR	Familial Male-Limited Precocious Puberty, Hypergonadotropic Hypogonadism	AD,AR	100%	75 of 75
LHX3	Sensorineural Deafness With Pituitary Dwarfism, Hypothyroidism, Non-Acquired Combined Pituitary Hormone Deficiency-Sensorineural Hearing Loss-Spine Abnormalities Syndrome	AR	99.97%	18 of 19
LHX4	Combined Pituitary Hormone Deficiencies, Hypothyroidism, Combined Pituitary Hormone Deficiency, Pituitary Stalk Interruption Syndrome	AD	99.95%	21 of 22
LHX8	Odontoma, Cerebral Hemisphere Lipoma		100%	2 of 2
LZTFL1	Bardet-Biedl Syndrome	AR	99.83%	4 of 4
MAP3K1	46XY Complete Gonadal Dysgenesis, 46XY Partial Gonadal Dysgenesis, 46XY Sex Reversal	AD	96.50%	31 of 32
MCM8	Premature Ovarian Failure, Natural At Age Menopause	AR	99.94%	10 of 10
MCM9	Ovarian Dysgenesis	AR	99.93%	12 of 12
MKKS	Bardet-Biedl Syndrome, Mckusick-Kaufman Syndrome	AR	89.96%	71 of 71
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome,meckel Syndrome, Type 1	AR	99.98%	49 of 49
MSH4	Fanconi Anemia Complementation Group A, Premature Ovarian Failure, Lynch Syndrome		99.70%	3 of 3
MTHFR	Homocystinuria Due To Deficiency Of N(5,10)-Methylene Tetrahydrofolate Reductase Activity, Isolated Anencephaly, Isolated Exencephaly, Neural Tube Defects, Schizophrenia, Thrombophilia Venous Thromboembolism	AD,AR	100%	122 of 122
MYO7A	Autosomal Dominant Sensorineural Deafness, Autosomal Recessive Neurosensory Deafness, Usher Syndrome Type 1, Type 2	AD,AR	100%	579 of 580
NANOS1	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Male Infertility With Teratozoospermia, Spermatogenic Failure	AD	75.55%	2 of 3
NANOS3	Freemartinism, Pediatric Germ Cell Cancer	-	99.99%	2 of 2

NOBOX	Premature Ovarian Failure	AD	90.55%	14 of 17
NPHP1	Bardet-Biedl Syndrome, Joubert Syndrome, Nephronophthisis, Senior-Loken Syndrome	AR	100%	58 of 59
NPHP3	Meckel Syndrome, Nephronophthisis, NPHP3-related Meckel-Like Syndrome, Renal-Hepatic-Pancreatic Dysplasia, Senior-Loken Syndrome	AR	99.99%	84 of 84
NR0B1	46XX Testicular Disorder Of Sex Development, 46XY Complete Gonadal Dysgenesis, 46XY Partial Gonadal Dysgenesis, Congenital Adrenal Hypoplasia	X,XR,G	99.87%	NA of NA
NR0B2	Obesity	AD,AR,MU,P	99.09%	15 of 15
NR5A1	46XX Gonadal Dysgenesis, 46XX Ovotesticular Disorder Of Sex Development, 46XX Sex Reversal, 46XX Testicular Disorder Of Sex Development, 46XY Complete Gonadal Dysgenesis, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Premature Ovarian Failure, Spermatogenic Failure	AD	99.97%	222 of 224
NSMF	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD	99.69%	11 of 11
OFD1	Joubert Syndrome, Orofaciodigital Syndrome Type 1, Type 6, Primary Ciliary Dyskinesia, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome Type 2	X,XR,XD,G	98.09%	NA of NA
PADI6	Preimplantation Embryonic Lethality	AR	na	na
PCSK1	Obesity Due To Prohormone Convertase I Deficiency	AR	99.98%	45 of 45
PGRMC1	Preterm Premature Ruputre of Membranes, Placental choriocarcinoma, Premature Ovarian Failure	-	100%	NA of NA
PHF6	Borjeson-Forssman-Lehmann Syndrome	X,XR,G	99.93%	NA of NA
PICK1	Spermatogenic Failure, TARP syndrome, Depression		100%	1 of 1
PLCZ1	Spermatogenic Failure	AR	99.78%	8 of 8
PNPLA6	Ataxia-Hypogonadism-Choroidal Dystrophy Syndrome, Autosomal Recessive Spastic Paraplegia Type, Boucher-Neuhauser Syndrome, Cerebellar Ataxia-Hypogonadism Syndrome, Laurence-Moon Syndrome, Oliver-Mcfarlane Syndrome, Autosomal Recessive Spastic Paraplegia	AR	100%	65 of 65
POF1B	Premature Ovarian Failure	X,XR,G	99.54%	NA of NA
POLR3B	Hypomyelination-Hypogonadotropic Hypogonadism-Hypodontia Syndrome, Hypomyelinating Leukodystrophy With Or Without Oligodontia And/Orhypogonadotropic Hypogonadism	AR	100%	61 of 61
POMC	Obesity Due To Pro-opiomelanocortin Deficiency	AD,AR,MU,P	99.98%	40 of 40
POU1F1	Combined Pituitary Hormone Deficiencies, Hypothyroidism Due To Deficient Transcription Factors Involved In Pituitary Development Or Function, Pituitary Hormone Deficiency	AD,AR	100%	43 of 44
PPARG	Berardinelli-Seip Congenital Lipodystrophy, Carotid Intimal Medial Thickness ,Noninsulin-Dependent Diabetes Mellitus, Lipodystrophy, Obesity,pparg-related Familial Partial Lipodystrophy	AD,AR,MU,P	99.94%	53 of 53
PROC	Severe Hereditary Thrombophilia Due To Congenital Protein C Deficiency	AD,AR	99.94%	406 of 406
PROK2	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD	100%	20 of 20
PROKR2	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism, Pituitary Stalk Interruption Syndrome, Septo-Optic Dysplasia Spectrum	AD	100%	64 of 64
PROM1	Cone Rod Dystrophy, Macular Dystrophy, Retinitis Pigmentosa, Stargardt Disease	AD,AR	99.61%	90 of 93
PROP1	Combined Pituitary Hormone Deficiencies, Hypothyroidism Due To Deficient Transcription Factors Involved In Pituitary Development Or Function, Non-Acquired Panhypopituitarism, Pituitary Dwarfism	AR	100%	35 of 36
PROS1	Severe Hereditary Thrombophilia Due To Congenital Protein S Deficiency, Thrombophilia Due To Protein S Deficiency	AD,AR	100%	403 of 405
PRPH2	Adult-Onset Foveomacular Vitelliform Dystrophy, Central Areolar Choroidal Dystrophy, Cone Rod Dystrophy, Retinitis Punctata Albescens, Vitelliform Macular Dystrophy, Patterned Dystrophy Of Retinal Pigment Epithelium, Retinitis Pigmentosa, Stargardt Disease	AD,AR	100%	188 of 188
PSMC3IP	46XX Gonadal Dysgenesis, Ovarian Dysgenesis	AR	99.96%	9 of 9
RDH5	Fundus Albipunctatus, Retinitis Punctata Albescens	AD,AR	100%	54 of 54
REC8	Azoospermia, 46XX Gonadal Dysgenesis		100%	4 of 4
RHO	Congenital Stationary Night Blindness, Fundus Albipunctatus, Retinitis Punctata Albescens, Retinitis Pigmentosa	AD,AR	100%	229 of 229
RLBP1	Bothnia Retinal Dystrophy, Fundus Albipunctatus, Retinitis Punctata Albescens, Rod-Cone Dystrophy, Retinitis Pigmentosa	AD,AR	100%	32 of 33
RPGRIP1L	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.96%	52 of 52
RSPO1	Palmoplantar Hyperkeratosis With Squamous Cell Carcinoma Of Skin And 46XX Sex Reversal, Palmoplantar Keratoderma-XX Sex Reversal-Predisposition To Squamous Cell Carcinoma Syndrome	AR	100%	6 of 7
SDCCAG8	Bardet-Biedl Syndrome, Senior-Loken Syndrome	AR	96.29%	18 of 19
SEMA3A	Brugada Syndrome, Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome	AD	100%	29 of 29
SEPTIN12	Spermatogenic Failure	AD	99.84%	5 of 5
SERPINC1	Antithrombin III Deficiency, Hereditary Thrombophilia Due To Congenital Antithrombin Deficiency	AD,AR	100%	400 of 407
SLC26A8	Spermatogenic Failure	AD	98.81%	5 of 5
SMC1B	Corneal Dystrophy, 46XX Gonadal Dysgenesis		99.22%	3 of 3
SOHLH1	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Ovarian Dysgenesis, Spermatogenic Failure	AD,AR	100%	9 of 10
SOX10	Kallmann Syndrome, Peripheral Demyelinating Neuropathy, Central Dysmyelinating Leukodystrophy, Waardenburg Syndrome, Hirschsprung Disease, Peripheral Demyelinating Neuropathy, Central Dysmyelination, Waardenburg Syndrome, Waardenburg-Shah Syndrome	AD	99.74%	139 of 147
SOX2	Anophthalmia/Microphthalmia-Esophageal Atresia Syndrome, Microphthalmia, Septo-Optic Dysplasia Spectrum	AD	99.91%	78 of 78
SOX3	46XX Testicular Disorder Of Sex Development, X-linked Mental Retardation With Isolated Growth Hormone Deficiency, Non-Acquired Panhypopituitarism, Septo-Optic Dysplasia Spectrum	X,G	92.88%	NA of NA

SOX9	46XX Ovotesticular Disorder Of Sex Development, 46XX Testicular Disorder Of Sex Development, 46XY Complete Gonadal Dysgenesis, 46XY Partial Gonadal Dysgenesis, Campomelic Dysplasia, Isolated Pierre Robin Syndrome	AD	97.28%	87 of 95
SPATA16	Spermatogenic Failure	AR	99.94%	1 of 2
SPRY4	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.72%	13 of 13
SRY	45X/ 46XY Mixed Gonadal Dysgenesis, 46XX Ovotesticular Disorder Of Sex Development, 46XX Testicular Disorder Of Sex Development, 46XY Complete Gonadal Dysgenesis, 46XY Partial Gonadal Dysgenesis, 46XX Sex Reversal, 46XY Sex Reversal	X,XD,Y,G	45%	NA of NA
STAG3	Premature Ovarian Failure	AR	98.88%	16 of 16
SULT2A1	Mixed Epithelial Stromal Tumor, Adrenal Cortical Adenoma, Polycystic Ovarian Syndrome, Adrenal Adenoma, Conn's Syndrome	-	99.97%	NA of NA
SUN5	Male Infertility Due To Acephalic Spermatozoa, Spermatogenic Failure	AR	100%	14 of 14
SYCE1	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Premature Ovarian Failure, Spermatogenic Failure	AR	100%	2 of 3
SYCP3	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AD	100%	5 of 5
TAC3	Hypogonadotropic Hypogonadism With Or Without Anosmia, Normosmic Congenital Hypogonadotropic Hypogonadism	AR	100%	10 of 10
TACR3	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AR	99.97%	40 of 40
TAF4B	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AR	97.92%	0 of 1
TEX11	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	X,XR,G	96.52%	NA of NA
TEX15	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AR	99.16%	6 of 7
THADA	Adenoma, Polycystic Ovary Syndrome	-	98.52%	NA of NA
TLE6	Preimplantation Embryonic Lethality	AR	100%	2 of 2
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome	AR	96.93%	177 of 179
TRIM32	Bardet-Biedl Syndrome, Limb-Girdle Muscular Dystrophy, TRIM 32-related Limb-Girdle Muscular Dystrophy	AR	100%	17 of 17
TTC21B	Asphyxiating Thoracic Dystrophy, Jeune Syndrome, Nephronophthisis, Joubert Syndrome	AD,AR	100%	67 of 67
TTC8	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	99.33%	28 of 28
TUBB8	Oocyte Maturation Defect	AD,AR	99.81%	47 of 47
USP9Y	Partial Chromosome Y Deletion, Y-linked Nonobstructive Spermatogenic Failure	Y,G	44.98%	NA of NA
WDPCP	Bardet-Biedl Syndrome, Congenital Heart Defects, Hamartomas Of Tongue And Polysyndactyly, Meckel Syndrome	AR	99.30%	8 of 8
WDR11	Hypogonadotropic Hypogonadism With Or Without Anosmia, Hypogonadotropic Hypogonadism Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism, Pituitary Stalk Interruption Syndrome	AD,AR	100%	19 of 19
WNT4	46XX Sex Reversal With Dysgenesis Of Kidneys, Adrenals, And Lungs, Mullerian Aplasia And Hyperandrogenism, Serkal Syndrome	AD,AR	100%	8 of 8
ZMYND15	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AR	99.87%	1 of 1
ZP1	Oocyte Maturation Defect	AR	100%	17 of 17
ZPBP	Spermatogenic Failure		99.98%	4 of 4

*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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