Premature Ovarian Insufficiency

Precision Panel

Overview

Premature Ovarian Insufficiency (POI) also known as premature ovarian failure or premature menopause, is defined as cessation of menstruation before the expected age of menopause. It is a loss of ovarian function before the age of 40 and it is a major cause of female infertility. There is an array of etiologies, including genetic, autoimmune and iatrogenic causes. It is a highly heterogeneous condition presenting with ovarian dysgenesis and primary amenorrhea, or with secondary amenorrhea and it can be associated with other congenital or acquired abnormalities. Since the age of menopause is an inheritable trait, POI has a strong genetic component worth investigating. POI is considered primary when the ovary fails to function normally and secondary if the hypothalamus and pituitary fail to provide appropriate gonadotropin stimulation. POI not only interferes with a woman's reproductive potential, but it is also associated with an increased risk of osteoporosis, cardiovascular disease and earlier mortality.

The Igenomix Premature Ovarian Insufficiency Precision Panel can be used to make a directed and accurate differential diagnosis of infertility ultimately leading to a better management of the patient 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 Premature Ovarian Insufficiency Precision Panel is indicated for those patients with clinical suspicion or diagnosis with or without the following manifestations:

- Signs of Turner syndrome: short stature, webbed neck, shieldlike chest etc
- Thyroid involvement: goiter, exophthalmos, bradycardia or tachycardia
- Adrenal insufficiency: orthostatic hypotension, hyperpigmentation, hypotension, decreased axillary and pubic hair
- Autoimmunity: vitiligo, nail dystrophy, mucocutaneous candidiasis, alopecia areata etc
- Hypoestrogenism: atrophic vaginitis, hot flashes, night sweats, vaginal dryness, irritability, decreased sexual desire
- Enlarged ovaries

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, workup and assisted reproductive technologies (ART), replacement hormonal therapy (RHT), and prevention of complications associated with multiorgan involvement.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

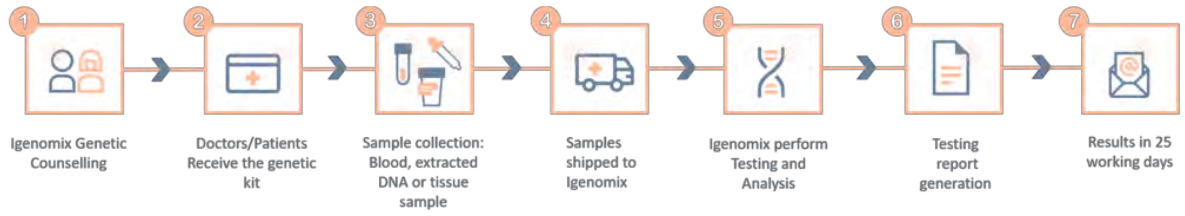
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AARS2	Combined Oxidative Phosphorylation Deficiency, Leukoencephalopathy, Progressive, With Ovarian Failure	AR	100	54 of 54
AIRE	Autoimmune Polyendocrinopathy Syndrome Type I	AD,AR	100	135 of 135
ANAPC1	Rothmund-Thomson Syndrome Type 1	AR	86.31	3 of 4
B4GALNT1	Autosomal Recessive Spastic Paraplegia	AR	98.69	13 of 13
BLM	Bloom Syndrome	AR	97.19	133 of 141
BMP15	Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	X,G	98.05	-
BNC1	Premature Ovarian Failure, 46,XX Gonadal Dysgenesis	AD	97.46	3 of 3
CASP10	Autoimmune Lymphoproliferative Syndrome	AD	99.86	6 of 6
CDH23	Pituitary Adenoma, Usher Syndrome, Cushing Disease	AD,AR	98	400 of 403
CEP164	Nephronophthisis, Senior-Loken Syndrome	AR	99.98	10 of 10
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CLPP	Perrault Syndrome	AR	99.91	11 of 11
CYP17A1	Congenital Adrenal Hyperplasia, 46,XY Disorder Of Sex Development Due To Isolated 17,20-Lyase Deficiency	AR	100	127 of 127
CYP19A1	Aromatase Deficiency , Aromatase Excess Syndrome	AD,AR	100	33 of 35
DCAF17	Woodhouse-Sakati Syndrome	AR	98.77	21 of 21
DIAPH2	Premature Ovarian Failure	X,XD,G	98.66	-
EIF2B1	Leukoencephalopathy With Vanishing White Matter	AR	100	9 of 9
EIF2B2	Leukoencephalopathy With Vanishing White Matter	AR	100	30 of 30
EIF2B3	Leukoencephalopathy With Vanishing White Matter	AR	97.55	26 of 26
EIF2B4	Leukoencephalopathy With Vanishing White Matter	AR	100	31 of 31
EIF2B5	Leukoencephalopathy With Vanishing White Matter	AR	100	99 of 99
ERAL1	Perrault Syndrome	AR	99.88	1 of 1
ERCC6	Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome, De Sanctis-Cacchione Syndrome, Premature Ovarian Failure	AD,AR	99.98	127 of 128
FANCM	Premature Ovarian Failure	AR	99.73	59 of 61
FAS	Autoimmune Lymphoproliferative Syndrome, Vogt-Koyanagi-Harada Disease	AD	100	135 of 135
FASLG	Autoimmune Lymphoproliferative Syndrome	AD	99.98	8 of 9
FIGLA	Premature Ovarian Failure	AD	98.47	4 of 5
FMR1	Fragile X Mental Retardation Syndrome, Premature Ovarian Failure, Xq27.3q28 Duplication Syndrome	X,XD,G	99.8	-
FOXL2	Premature Ovarian Failure	AD	89.36	136 of 201
FSHB	Hypogonadotropic Hypogonadism Without Anosmia, Isolated Follicle Stimulating Hormone Deficiency	AR	100	8 of 8
FSHR	Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AD,AR	100	41 of 43
GALT	Classic Galactosemia	AR	100	350 of 350
GDF9	Premature Ovarian Failure	AR	100	13 of 13

GNAS	ACTH-Independent Macronodular Adrenal Hyperplasia, McCune-Albright Syndrome	AD	99.95	263 of 273
GNRHR	Hypogonadotropic Hypogonadism Without Anosmia, Normosmic Congenital Hypogonadotropic Hypogonadism	AR	100	59 of 59
HFM1	Premature Ovarian Failure	AR	99.17	10 of 10
INVS	Senior-Loken Syndrome	AR	99.9	38 of 38
IQCB1	Senior-Loken Syndrome	AR	99.98	43 of 43
KISS1R	Hypogonadotropic Hypogonadism With Or Without Anosmia, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism	AD,AR	99.41	42 of 43
LARS2	Perrault Syndrome	AR	99.99	20 of 20
LHB	Hypogonadotropic Hypogonadism Without Anosmia	AR	100	11 of 11
LHCGR	Hypergonadotropic Hypogonadism	AD,AR	100	75 of 75
LMNA	Dilated Cardiomyopathy-Hypergonadotropic Hypogonadism Syndrome	AD,AR	100	619 of 620
MCM3AP	Autosomal Recessive Peripheral Neuropathy With Or Without Impaired Intellectual Development	AR	99.96	22 of 22
MCM8	Premature Ovarian Failure	AR	99.94	10 of 10
MCM9	Ovarian Dysgenesis	AR	99.93	12 of 12
MRPS22	Combined Oxidative Phosphorylation Deficiency, Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	100	10 of 10
MSH5	Premature Ovarian Failure	AR	100	4 of 4
NBN	Nijmegen Breakage Syndrome, Hereditary Breast And Ovarian Cancer Syndrome	AR,MU,P	100	200 of 200
NOBOX	Premature Ovarian Failure	AD	90.55	14 of 17
NPHP1	Joubert Syndrome, Senior-Loken Syndrome, Bardet-Biedl Syndrome	AR	100	58 of 59
NPHP3	Meckel Syndrome, Senior-Loken Syndrome	AR	99.99	84 of 84
NPHP4	Senior-Loken Syndrome	AR	99.96	118 of 119
NR5A1	46,XX Sex Reversal, 46,XY Sex Reversal, Premature Ovarian Failure, 46,XX Gonadal Dysgenesis	AD	99.97	222 of 224
NUP107	Galloway-Mowat Syndrome, Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	99.91	15 of 15
PMM2	Congenital Disorder Of Glycosylation	AR	100	127 of 129
POF1B	Premature Ovarian Failure	X,XR,G	99.54	-
POLG	Mitochondrial DNA Depletion Syndrome	AD,AR	99.92	325 of 326
POLR3H	46,XX Gonadal Dysgenesis	-	99.96	1 of 1
POR	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis	AD,AR	99.98	67 of 68
PRKCD	Autoimmune Lymphoproliferative Syndrome	AR	100	9 of 9
PSMC3IP	Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	99.96	9 of 9
RASGRP1	Autoimmune Lymphoproliferative Syndrome	AR	98.41	8 of 9
RCBTB1	Retinal Dystrophy With Or Without Extraocular Anomalies	AR	99.94	10 of 10
RIN2	Macrocephaly, Alopecia, Cutis Laxa, And Scoliosis, Rin2 Syndrome	AR	99.6	4 of 4
SDCCAG8	Bardet-Biedl Syndrome, Senior-Loken Syndrome	AR	96.29	18 of 19
SOHLH1	Ovarian Dysgenesis	AD,AR	100	9 of 10
SPIDR	46,XX Gonadal Dysgenesis	-	82	1 of 1
STAG3	Premature Ovarian Failure	AR	98.88	16 of 16
STAR	Lipoid Congenital Adrenal Hyperplasia, Familial Glucocorticoid Deficiency	AR	100	80 of 80
SYCE1	Premature Ovarian Failure	AR	100	2 of 3
THOC6	Microcephaly, Mental Retardation, And Distinctive Facies, With Cardiacand Genitourinary Malformations	AR	100	13 of 13
TRAF3IP1	Senior-Loken Syndrome	AR	97.54	15 of 15
TTI2	Severe Intellectual Disability-Short Stature-Behavioral Abnormalities-Facial Dysmorphism Syndrome	AR	100	6 of 6
TWNK	Perrault Syndrome, Progressive External Ophthalmoplegia With Mitochondrial DNA Deletions	AD,AR	-	-
USP8	ACTH-Secreting Pituitary Adenoma, Cushing Disease	AD,AR	98.19	3 of 3
WDR19	Senior-Loken Syndrome, Jeune Syndrome	AR	99.96	47 of 49
WT1	Denys-Drash Syndrome, Frasier Syndrome, 46,XY Complete Gonadal Dysgenesis , 46,XY Partial Gonadal Dysgenesis, Denys-Drash Syndrome, Wagr Syndrome	AD	98.92	178 of 185

*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.

References

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