Recurrent Pregnancy Loss

Precision Panel

Overview

Recurrent Pregnancy Loss (RPL) is one of the most common obstetric complications, affecting more than 30% of conceptions. These can occur during preimplantation, pre-embryonic, embryonic, early fetal, late fetal and stillbirth. An important number of losses are due to genetic abnormalities, nonetheless 50% of early pregnancy losses have been associated with chromosomal abnormalities. The majority are due to de novo non-disjunctional events during meiosis and balanced paternal translocations. Traditionally, the assessment of recurrent pregnancy loss was based on karyotyping techniques. However, advances in molecular genetic technology have provided an array of information regarding genetic causes and risk factors for pregnancy loss. One of the most innovative techniques with a significant role in RPL is preimplantation genetic testing in in vitro fertilization cycles.

The Igenomix Recurrent Pregnancy Loss Precision Panel can be used to make a directed and accurate differential diagnosis of inability to carry out a full pregnancy 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 Recurrent Pregnancy Loss Precision Panel is indicated for those patients the following manifestations:

- Inability to conceive after 1 year of unprotected intercourse
- Family history of infertility
- Personal history of recurrent miscarriages
- Family history of recurrent miscarriages
- Previous failed IVF cycles
- Other failed assisted reproductive technology (ART) treatments

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).

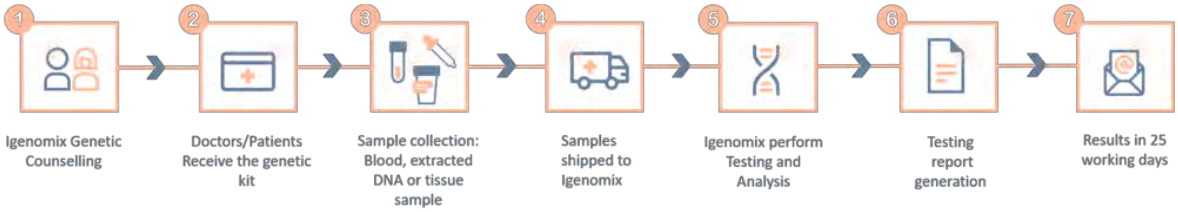
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
CTNNA3	Familial Arrhythmogenic Right Ventricular Dysplasia	AD	99.97%	14 of 17
DYNC2H1	Jeune Syndrome, Short Rib-Polydactyly Syndrome, Verma-Naumoff Type, Short-Rib Thoracic Dysplasia With Or Without Polydactyly	AR,MU,D	99.78%	214 of 221
F2	Congenital Factor II Deficiency, Congenital Prothrombin Deficiency, Ischemic Stroke, Recurrent Pregnancy Loss, Venous Thromboembolism	AD,AR,MU	100%	66 of 66
GLE1	Amyotrophic Lateral Sclerosis, Congenital Arthrogryposis With Anterior Horn Cell Disease, Lethal Congenital Contracture Syndrome	AR	100%	17 of 17
ITGB3	Fetal And Neonatal Alloimmune Thrombocytopenia, Glanzmann Thrombasthenia	AD,AR	99.44%	178 of 179
KCNH2	Familial Short QT Syndrome, Long Qt Syndrome, Romano-Ward Syndrome	AD	98.69%	908 of 930
KCNQ1	Familial Atrial Fibrillation, Beckwith-Wiedemann Syndrome, Familial Short QT Syndrome, Jervell And Lange-Nielsen Syndrome, Long QT Syndrome, Romano-Ward Syndrome	AD,AR	93.23%	600 of 624
KIF14	Autosomal Recessive Primary Microcephaly, Meckel Syndrome, Primary Microcephaly	AR	99.84%	18 of 18
MECP2	Atypical Rett Syndrome, X-linked Autism, Severe Neonatal Encephalopathy Due To MECP2 Mutations, Lubs X-linked Mental Retardation Syndrome, Rett Syndrome, Trisomy Xq28,x-Linked Intellectual Disability-Psychosis-Macroorchidism Syndrome, X-linked Non-Syndromic Intellectual Disability	X,XR,XD,MU,G	99.81%	NA of NA
MTHFR	Homocystinuria Due To Deficiency Of N(5,10)-Methylene Tetrahydrofolate Reductase Activity, Isolated Anencephaly, Isolated Exencephaly, Neural Tube Defects, Folate-Sensitive, Schizophrenia, Venous Thromboembolism	AD,AR	100%	122 of 122
RYR1	Autosomal Dominant Centronuclear Myopathy, Autosomal Recessive Centronuclear Myopathy, Benign Samaritan Congenital Myopathy, Central Core Disease, Congenital Multicore Myopathy With External Ophthalmoplegia, Congenital Myopathy With Myasthenic-like Onset, Malignant Hyperthermia Of Anesthesia	AD,AR	97.63%	733 of 746
SCN5A	Familial Atrial Fibrillation, Brugada Syndrome, Dilated Cardiomyopathy, Familial Progressive Cardiac Conduction Defect, Long QT Syndrome, Progressive Familial Heart Block, Romano-Ward Syndrome, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation	AD,AR,MU	99.45%	929 of 942
SERPINE1	Plasminogen Activator Inhibitor-1 Deficiency	AD,AR	100%	4 of 4
TIMP2	Conjuctivochalasis, Sorsby Fundus Dystrophy, Fibrosarcoma, Preterm Premature Rupture of the Membranes		97.56%	6 of 6

*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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