Embryo Developmental Arrest

Precision Panel

Overview

Embryo Developmental Arrest (EDA) is one of the mechanisms responsible for an increased level of embryo demise during the first week of *in vitro* development. Around 10-15% embryos permanently arrest in mitosis at the 2-to 4-cell cleavage stage. It involves the downregulation and/or cessation of cell division and metabolic activity of the components involved in the formation and development of an embryo. Chromosomal abnormalities, abnormal preimplantation development and single gene disorders have been stated as causes of EDA and therefore, a known cause of infertility. The identification of abnormal gene changes previously known to have an effect on embryo development is crucial to improve pregnancy outcomes.

The Igenomix Embryo Developmental Arrest 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 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
- Personal or 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.

GPDx Genomic Precision Diagnostic by Igenomix

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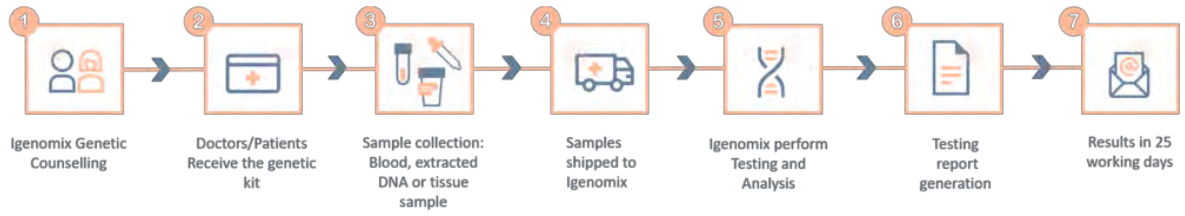
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
BTG4	Zygotic Cleavage Failure, Female Infertility		92.22%	NA of NA
C1QC	C1q Deficiency	AR	100%	11 of 11
CATSPER1	Spermatogenic Failure	AR	99.97%	4 of 4
CD46	HELLP Syndrome, Hemolytic Uremic Syndrome	AD,AR	100%	83 of 84
CNOT6L	Embryo Developmental Arrest	-	98.92%	NA of NA
DMC1	Infertility	-	100%	2 of 2
DNAH1	Primary Ciliary Dyskinesia, Spermatogenic Failure	AR	100%	58 of 58
DNAH5	Primary Ciliary Dyskinesia With Or Without Situs Inversus	AR	100%	277 of 278
DPY19L2	Spermatogenic Failure	AR	97.65%	16 of 20
EED	Cohen-Gibson Syndrome, Weaver Syndrome	AD	99.92%	10 of 10
GALNTL5	Primary Infertility Due to Asthenozoospermia		99.95%	2 of 2
KHDC3L	Recurrent Hydatidiform Mole, Recurrent	AR	100%	7 of 7
KLHL10	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AD	99.98%	5 of 5
NANOG	Teratocarcinoma, Germ Cell and Embryonal Cancer		97.74%	NA of NA
NANOS1	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Male Infertility With Teratozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AD	75.55%	2 of 3
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
PADI6	Preimplantation Embryonic Lethality	AR	NA	NA
PICK1	Spermatogenic Failure, Depression		100%	1 of 1
PLCZ1	Spermatogenic Failure	AR	99.78%	8 of 8
POU5F1	Embryonal Carcinoma, Teratoma		100%	1 of 1
SEPTIN12	Spermatogenic Failure	AD	99.84%	5 of 5
SLC26A8	Spermatogenic Failure	AD	98.81%	5 of 5
SPATA16	Spermatogenic Failure	AR	99.94%	1 of 2
SPP1	Pediatric Systemic Lupus Erythematosus		99.77%	2 of 2
STAT3	Acute Promyelocytic Leukemia, Infantile-Onset Autoimmune Disease, Hyper-IgE Syndrome, Permanent Neonatal Diabetes Mellitus	AD	100%	171 of 171
STK11	Pancreatic Cancer, Peutz-Jeghers Syndrome,Testicular Tumor	AD	81.99%	456 of 470
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
TAF4B	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Spermatogenic Failure	AR	97.92%	0 of 1
TERT	Aplastic Anemia, Dyskeratosis Congenita, Familial Melanoma, Hoyeraal-Hreidarsson Syndrome, Idiopathic Aplastic Anemia, Idiopathic Pulmonary Fibrosis, Acute Myeloid Leukemia, Cutaneous Malignant Melanoma, Meningioma, Pulmonary Fibrosis And/Or Bone Marrow Failure, Telomere-related, Pulmonary Fibrosis	AD,AR	99.09%	194 of 197
TEX11	Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, X-linked 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
TLE6	Preimplantation Embryonic Lethality	AR	100%	2 of 2
TUBB8	Oocyte Maturation Defect	AD,AR	99.81%	47 of 47
VSIG4	T-cell/Histiocyte Rich Large B Cell Lymphoma, Complement Component 3 Deficiency, Hemolytic Uremic Syndrome	-	99.80%	NA of NA
ZFP42	Spermatocytoma, Germ Cell and Embryonal Cancer	-	99.98%	NA of NA
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.

References

1. Murphy, B. (2020). Under Arrest: The Embryo in Diapause. *Developmental Cell*, *52*(2), 139-140. doi: 10.1016/j.devcel.2020.01.002
2. Levy, R. R., Cordonier, H., Czyba, J. C., & Guerin, J. F. (2001). Apoptosis in preimplantation mammalian embryo and genetics. *Italian journal of anatomy and embryology = Archivio italiano di anatomia ed embriologia*, *106*(2 Suppl 2), 101–108.
3. Mohebi, M., & Ghafouri-Fard, S. (2019). Embryo developmental arrest: Review of genetic factors and pathways. *Gene Reports*, *17*, 100479. doi: 10.1016/j.genrep.2019.100479
4. Zhang, X., Stojkovic, P., Przyborski, S., Cooke, M., Armstrong, L., Lako, M., & Stojkovic, M. (2006). Derivation of Human Embryonic Stem Cells from Developing and Arrested Embryos. *Stem Cells*, *24*(12), 2669-2676. doi: 10.1634/stemcells.2006-0377
5. Sha, Q. Q., Zheng, W., Wu, Y. W., Li, S., Guo, L., Zhang, S., Lin, G., Ou, X. H., & Fan, H. Y. (2020). Dynamics and clinical relevance of maternal mRNA clearance during the oocyte-to-embryo transition in humans. *Nature communications*, *11*(1), 4917. https://doi.org/10.1038/s41467-020-18680-6
6. Zhang, Y., Feng, Y., & Ma, F. (2020). *Yi chuan = Hereditas*, *42*(10), 1004–1016. https://doi.org/10.16288/j.yczz.20-144
7. Feng, R., Yan, Z., Li, B., Yu, M., Sang, Q., Tian, G., Xu, Y., Chen, B., Qu, R., Sun, Z., Sun, X., Jin, L., He, L., Kuang, Y., Cowan, N. J., & Wang, L. (2016). Mutations in TUBB8 cause a multiplicity of phenotypes in human oocytes and early embryos. *Journal of medical genetics*, *53*(10), 662–671. https://doi.org/10.1136/jmedgenet-2016-103891
8. Xu, Y., Shi, Y., Fu, J., Yu, M., Feng, R., & Sang, Q. et al. (2016). Mutations in PADI6 Cause Female Infertility Characterized by Early Embryonic Arrest. *The American Journal Of Human Genetics*, *99*(3), 744-752. doi: 10.1016/j.ajhg.2016.06.024