Gonadal Dysgenesis

Precision Panel

Overview

Gonadal Dygenesis comprises a clinical spectrum of anomalies in patients with female, ambiguous or male phenotype, absent or impaired puberty and karyotype with or without Y chromosome. It is usually defined as congenital hypogonadism related to abnormalities of the sex chromosomes. The identification of dysgenetic gonads is crucial because they are potentially prone to developing tumors such as gonadoblastoma. The most notable of these conditions is Turner syndrome, with an array of associated symptoms and complications.

The Igenomix Gonadal Dysgenesis 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 Gonadal Dysgenesis Precision Panel is indicated for those patients with clinical suspicion or diagnosis with or without the following manifestations:

- Short stature
- Primary amenorrhea
- Streak gonads
- Sexual infantilism
- Ultrasound-karyotype discordance of genotype
- Failure to develop secondary sex characteristics

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, surgical repair, assisted reproductive technologies (ART), hormone replacement therapy and surveillance for neoplasms.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

- Understanding the genetics behind gonadal dysgenesis allowing clinicians to better predict the disorder's phenotypic presentation, improving screening methods and ongoing care of those medical problems.

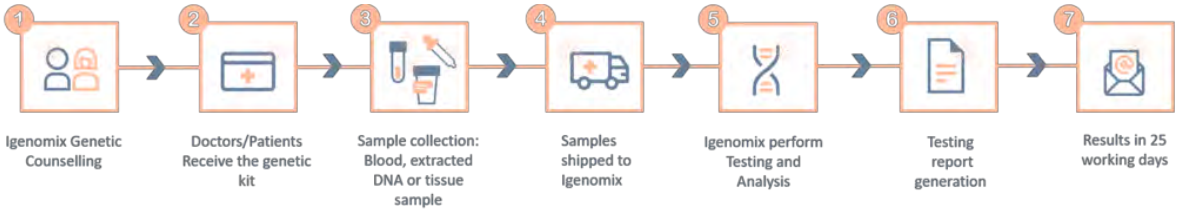
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
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
CBX2	46XY Sex Reversal, 46,XY Complete Gonadal Dysgenesis	AR	100	6 of 6
DHH	46,XY Gonadal Dysgenesis, 46,XY Sex Reversal, 46,XY Gonadal Dysgenesis-Motor And Sensory Neuropathy Syndrome	AR	99.85	21 of 21
DHX37	46,XY Sex Reversal, Neurodevelopmental Disorder With Brain Anomalies And With Or Without Vertebral Or Cardiac Anomalies, 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis, Testicular Regression Syndrome	AD,AR	99.87	13 of 13
DMRT1	46,XY Complete Gonadal Dysgenesis	-	99.93	6 of 7
DMRT3	46,XY Partial Gonadal Dysgenesis	-	88.67	1 of 1
ERCC2	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	100	102 of 102
ERCC3	Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.98	24 of 24
FSHR	Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AD,AR	100	41 of 43
GATA4	Testicular Anomalies With Or Without Congenital Heart Disease, 46,XY Partial Gonadal Dysgenesis	AD	94.69	108 of 130
GTF2E2	Nonphotosensitive Trichothiodystrophy	AR	99.98	2 of 2
GTF2H5	Photosensitive Trichothiodystrophy	AR	100	8 of 8
HSD17B4	D-Bifunctional Protein Deficiency, Perrault Syndrome	AR	99.52	85 of 85
MAP3K1	46,XY Sex Reversal, 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis	AD	96.5	31 of 32
MPLKIP	Nonphotosensitive Trichothiodystrophy	AR	100	13 of 13
MRPS22	Combined Oxidative Phosphorylation Deficiency, Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	100	10 of 10
NR0B1	Congenital Adrenal Hypoplasia, Dosage-Sensitive Sex Reversal , 46,XX Testicular Disorder Of Sex Development , 46,XY Complete Gonadal Dysgenesis , 46,XY Partial Gonadal Dysgenesis	X,XR,G	99.87	-
NR5A1	46,XX Sex Reversal, 46,XY Sex Reversal, Premature Ovarian Failure, Spermatogenic Failure, 46,XX Gonadal Dysgenesis, 46,XX Ovotesticular Disorder Of Sex Development , 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis	AD	99.97	222 of 224
NUP107	Galloway-Mowat Syndrome, Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	99.91	15 of 15
POLR3H	46,XX Gonadal Dysgenesis		99.96	1 of 1
PPP1R12A	Genitourinary And/Or/Brain Malformation Syndrome	AD	99.48	1 of 1
PPP2R3C	Gonadal Dysgenesis, Dysmorphic Facies, Retinal Dystrophy, And Myopathy	AD,AR	99.85	3 of 3
PSMC3IP	Ovarian Dysgenesis, 46,XX Gonadal Dysgenesis	AR	99.96	9 of 9
RNF113A	Nonphotosensitive Trichothiodystrophy	X,XD,G	99.7	-
RXYLT1	Walker-Warburg Syndrome	AR	99.46	-
SOX9	46,XX Ovotesticular Disorder Of Sex Development, 46,XY Complete Gonadal Dysgenesis , 46,XY Partial Gonadal Dysgenesis	AD	97.28	87 of 95
SPIDR	46,XX Gonadal Dysgenesis	-	82	1 of 1
SRY	46,XX Sex Reversal, 46XY Sex Reversal, 45,x/46,XY Mixed Gonadal Dysgenesis, 46,XX Ovotesticular Disorder Of Sex Development, 46,XY Complete Gonadal Dysgenesis , 46,XY Partial Gonadal Dysgenesis	X,XD,Y,G	45	-
TARS1	Nonphotosensitive Trichothiodystrophy	AR	99.94	-
TOE1	Pontocerebellar Hypoplasia	AR	99.98	12 of 12
TWNK	Infantile-Onset Spinocerebellar Ataxia, Perrault Syndrome	AD,AR	-	-
VAMP7	46,XY Partial Gonadal Dysgenesis	-	99.98	-
WT1	Denys-Drash Syndrome, Frasier Syndrome, 46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis, Meacham Syndrome	AD	98.92	178 of 185
WWOX	Early Infantile Epileptic Encephalopathy, Spinocerebellar Ataxia, 46,XY Partial Gonadal Dysgenesis	AR	99.94	44 of 44

ZFPM2	46,XY Sex Reversal, 46,XY Partial Gonadal Dysgenesis	AD	99.4	44 of 46

*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. Lipay, M. V., Bianco, B., & Verreschi, I. T. (2005). Disgenesias gonadais e tumores: aspectos genéticos e clínicos [Gonadal dysgenesis and tumors: genetic and clinical features]. *Arquivos brasileiros de endocrinologia e metabologia*, *49*(1), 60–70. https://doi.org/10.1590/s0004-27302005000100008
2. Breuil, V., & Euller-Ziegler, L. (2001). Gonadal dysgenesis and bone metabolism. *Joint bone spine*, *68*(1), 26–33. https://doi.org/10.1016/s1297-319x(00)00235-9
3. XY gonadal dysgenesis. (1979). *Lancet (London, England)*, *1*(8106), 27.
4. Breehl L, Caban O. Genetics, Gonadal Dysgenesis. [Updated 2020 Oct 3]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2021 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK539886/
5. McDonough, P. G., & Byrd, J. R. (1977). Gonadal dysgenesis. *Clinical obstetrics and gynecology*, *20*(3), 565–579. https://doi.org/10.1097/00003081-197709000-00007
6. Ferguson-Smith M. A. (1965). Karyotype-Phenotype Correlations In Gonadal Dysgenesis And Their Bearing On The Pathogenesis Of Malformations. *Journal of medical genetics*, *2*(2), 142–155. https://doi.org/10.1136/jmg.2.2.142