DiGeorge Syndrome
Precision Panel

Overview

DiGeorge Syndrome (DGS) is one of a group of phenotypically similar disorders including velocardiofacial syndrome (VCFS) and conotruncal anomaly face (CTAF). These disorders share a microdeletion of chromosome 22q11.2, a region known as the DiGeorge critical region. Since there are overlapping phenotypic features between these syndromes, they have been designated as 22q11.2 deletion syndromes. It is one of the most common chromosomal microdeletion disorders. The 22q11.2 deletion results in a range of embryonic developmental disruptions involving the head, nek, brain, skeleton, and kidneys. There is also a defect in T cell production which in turn increases susceptibility for infections as well autoimmunity. Neuropsychiatric manifestations of these disorders cause moderate/severe impairment of functionality in these patients. The prognosis for these disorders is highly variable, depending on the nature and degree of involvement of different organs.

The Igenomix DiGeorge Syndrome Precision Panel can serve as a directed and accurate diagnostic tool ultimately leading to a better management and prognosis of the disease. 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.

Indications

The Igenomix DiGeorge Syndrome Precision Panel is indicated in those cases where there is a clinical suspicion or diagnosis with or without the following manifestations:

- Retrognathia or micrognathia
- Long face
- High and broad nasal bridge
- Narrow palpebral fissures
- Small teeth
- Short philtrum
- Low-set, malformed ears
- Congenital heart defects
- Hypocalcemia
- Cognitive, behavioral and psychiatric problems
- Increased susceptibility to infections

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis and improve prognosis.

- Early initiation of treatment with a multidisciplinary approach involving pediatrics, general medicine, surgery, psychiatry, psychology, interventional therapies (physical, occupational, speech, language and behavioral).
- Start early medical care with calcium supplementation, surgical care and bone marrow transplantation for profound immunodeficiency, early surveillance and prevention of complications.
- Risk assessment and genetic counselling of asymptomatic family members to identify the individuals at risk.
- Improvement of delineation of genotype-phenotype correlation.
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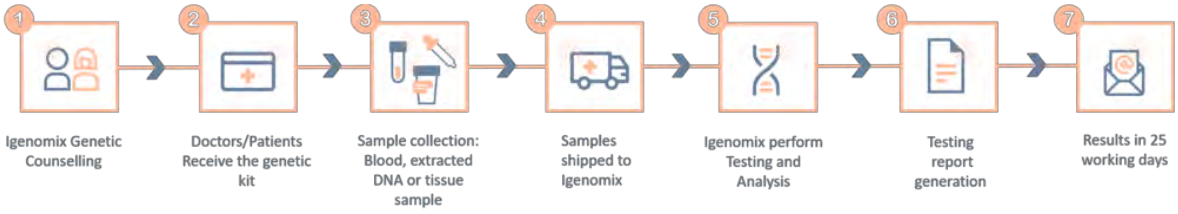
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ARVCF	22q11.2 Deletion Syndrome	-	99.95	2 of 2
COMT	Panic Disorder, Schizophrenia, 22q11.2 Deletion Syndrome	AD	99.98	5 of 5
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Thrombocytopenia	AR	74.08	26 of 50
HIRA	22q11.2 Deletion Syndrome	-	99.99	5 of 5
JMJD1C	22q11.2 Deletion Syndrome	-	99.09	27 of 27
RREB1	22q11.2 Deletion Syndrome	-	99.92	8 of 8
SEC24C	22q11.2 Deletion Syndrome	-	99.98	-
TBX1	Conotruncal Heart Malformations, Digeorge Syndrome, Tetralogy Of Fallot, Velocardiofacial Syndrome, 22q11.2 Deletion Syndrome, 22q11.2 Microduplication Syndrome	AD,AR	88.7	35 of 42
UFD1	22q11.2 Deletion Syndrome	-	99.98	-

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: 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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