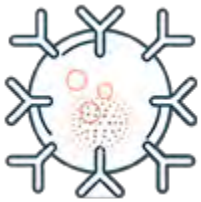
Prader Willi/Angelman Syndrome

Precision Panel



Overview

Prader Willi and Angelman Syndrome are neurodevelopmental disorders caused by a deletion of a region in chromosome 15 and are classically known as genomic imprinting disorders. These disorders are characterized by an imprinting center controlling the expression of selected genes in chromosome 15, therefore a deletion in these areas affect the expression of certain genes. Depending on the deletion occurring in the maternal or paternal chromosome the resulting disorder is Prader Willi if occurring in the paternal chromosome, or Angelman Syndrome if occurring in the maternal chromosome. Prader Willi Syndrome is caused by paternal deletion or maternal uniparental disomy of chromosome 15 and it is commonly characterized by diminished fetal activity, obesity, hypotonia, intellectual disability, short stature etc. Angelman Syndrome is originated from maternal deletion or paternal uniparental disomy and is characterized by jerky movements, abnormal laugher, sleep disturbances and characteristic facial features.

The Igenomix Prader Willi and Angelman Syndrome Precision Panel can be used for an accurate and directed diagnosis 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 involved.

Indications

The Igenomix Prader Willi and Angelman Syndrome Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Hypotonia
- Feeding problems or failure to thrive
- Rapid weight gain
- Facial features: Narrow bifrontal diameter, almond-shaped palpebral fissures, narrow nasal bridge
- Hypogonadism
- Developmental delay and severe intellectual deficit
- Microcephaly
- Speech impairment
- Epilepsy
- Tongue protrusion
- Paroxysms of laughter
- Abnormal sleep patterns
- Hyperactivity

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 involving a multidisciplinary team in the form management of hypotonia and poor feeding, evaluation of hypogonadism, management of obesity, monitoring for scoliosis and therapy for behavioral issues and surgical care if needed.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
HERC2	Autosomal Recessive Mental Retardation, Prader-Willi Syndrome	AD,AR	98.91	9 of 9
IPW	Prader-Willi Syndrome	AD	-	-
MAGEL2	Prader-Willi Syndrome	AD	99.99	43 of 48
MBD5	Autosomal Dominant Mental Retardation, 2q23.1 Microdeletion Syndrome	AD	99.99	33 of 35
MKRN3	Prader-Willi Syndrome	AD,AD,MI	99.98	39 of 41
MKRN3-AS1	Prader-Willi Syndrome	AD	-	-
NDN	Prader-Willi Syndrome	AD	97.41	2 of 2
NPAP1	Prader-Willi Syndrome	AD	99.82	-
PWAR1	Prader-Willi Syndrome	AD	-	-
PWRN1	Prader-Willi Syndrome ,	AD	-	-
SIM1	6q16 Microdeletion Syndrome , Obesity, Prader-Willi-like Syndrome	-	99.64	39 of 40
SLC9A6	X-linked Syndromic Mental Retardation, Christianson Syndrome	X,XD,G	98.87	-
SNORD115-1	Prader-Willi Syndrome	AD	-	-
SNORD116-1	Prader-Willi Syndrome	AD	-	-
SNRPN	Prader-Willi Syndrome, Autism	AD,MU	100	2 of 2
UBE3A	Angelman Syndrome , 15q11q13 Microduplication Syndrome	AD	99.98	208 of 211

*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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