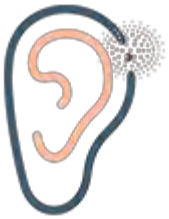
Waardenburg Syndrome

Precision Panel



Overview

Waardenburg Syndrome (WS) is a genetic disorder characterized by the association of pigmentation abnormalities, including depigmented patches of the skin and hair, blue eyes (heterochromia irides), and sensorineural hearing loss. It also presents with other clinical features involving musculoskeletal abnormalities, gastrointestinal malformations and neurological defects. WS is considered a defect in the melanocyte and neural crest development, where a complex interconnecting regulatory network of genes work in synergism for an appropriate development of melanocytes. It is typically inherited in an autosomal dominant pattern.

The Igenomix Waardenburg Syndrome Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of hearing loss 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 Waardenburg Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Cleft lip
- Constipation
- Deafness
- Extremely pale blue eyes or nonmatching eye colors
- Pale color skin, hair and eyes
- Decreased intellectual function

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 in the form of hearing aids and cochlear implants, social services and speech therapy.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

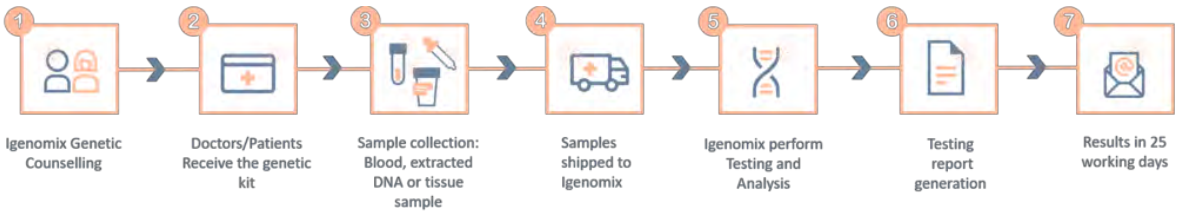
- Improvement of delineation of genotype-phenotype correlation.
- Unravel important developmental pathways in the neural crest and derivatives that could potentially lead to Waardenburg Syndrome.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
EDN3	Congenital Failure Of Autonomic Control, Waardenburg Syndrome, Ondine Syndrome, Waardenburg-Shah Syndrome	AD,AR	100	20 of 22
EDNRB	Abcd Syndrome, Waardenburg-Shah Syndrome, Waardenburg Syndrome	AD,AR	99.55	70 of 72
KIT	Gastrointestinal Stromal Tumor, Acute Myeloid Leukemia, Mast Cell Disease, Piebald Trait	AD	100	112 of 112
KITLG	Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome	AD	99.93	10 of 10
MITF	Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, And Deafness, Tietz Syndrome, Waardenburg Syndrome, Waardenburg-Shah Syndrome	AD,AR	100	72 of 72
PAX3	Craniofacial-Deafness-Hand Syndrome, Alveolar Rhabdomyosarcoma, Waardenburg Syndrome, Craniofacial-Deafness-Hand Syndrome	AD,AR	99.98	157 of 157
SMOC1	Microphthalmia With Limb Anomalies	AR	100	19 of 19
SNAI2	Piebald Trait, Waardenburg Syndrome	AD,AR	99.79	1 of 2
SOX10	Peripheral Demyelinating Neuropathy, Central Dysmyelination, Waardenburg Syndrome, Waardenburg-Shah Syndrome	AD	99.74	139 of 147

*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. Pingault, V., Ente, D., Dastot-Le Moal, F., Goossens, M., Marlin, S., & Bondurand, N. (2010). Review and update of mutations causing Waardenburg syndrome. *Human mutation*, *31*(4), 391–406. https://doi.org/10.1002/humu.21211

2. Saleem M. D. (2019). Biology of human melanocyte development, Piebaldism, and Waardenburg syndrome. *Pediatric dermatology*, *36*(1), 72–84. https://doi.org/10.1111/pde.13713
3. Ren, S., Chen, X., Kong, X., Chen, Y., Wu, Q., Jiao, Z., & Shi, H. (2020). Identification of six novel variants in Waardenburg syndrome type II by next-generation sequencing. *Molecular Genetics & Genomic Medicine*, *8*(3). doi: 10.1002/mgg3.1128
4. Read, A. P., & Newton, V. E. (1997). Waardenburg syndrome. *Journal of medical genetics*, *34*(8), 656–665. https://doi.org/10.1136/jmg.34.8.656
5. Tagra, S., Talwar, A. K., Walia, R. L., & Sidhu, P. (2006). Waardenburg syndrome. *Indian journal of dermatology, venereology and leprology*, *72*(4), 326. https://doi.org/10.4103/0378-6323.26718
6. Ringer J. (2019). Identification of Waardenburg Syndrome and the Management of Hearing Loss and Associated Sequelae: A Review for the Pediatric Nurse Practitioner. *Journal of pediatric health care : official publication of National Association of Pediatric Nurse Associates & Practitioners*, *33*(6), 694–701. https://doi.org/10.1016/j.pedhc.2019.06.001
7. Minami, S. B., Nara, K., Mutai, H., Morimoto, N., Sakamoto, H., Takiguchi, T., Kaga, K., & Matsunaga, T. (2019). A clinical and genetic study of 16 Japanese families with Waardenburg syndrome. *Gene*, *704*, 86–90. https://doi.org/10.1016/j.gene.2019.04.023