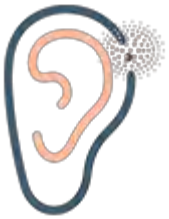
Usher Syndrome

Precision Panel



Overview

Usher Syndrome (USH) is a clinically and genetically heterogeneous disorder presenting with sensorineural hearing loss due to an impaired ability of the inner ear and auditory nerves to transmit sensory input to the brain. It is usually accompanied by vestibular involvement and retinitis pigmentosa which is characterized by progressive loss of vision. It is the leading genetic cause of combined hearing and vision loss. Based on the hearing and vestibular symptoms it has been classified into three types: type 1 is the most severe form; type 2 is the most frequent form and type 3 is the rarest and most heterogeneous form. It is transmitted in an autosomal recessive manner.

The Igenomix Usher Syndrome Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of sensorineural 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 Usher Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of Usher Syndrome presenting with:

- Deafness
- Balance problems
- Night blindness
- Family history of deafness, balance problems and night blindness

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.

GPDx Genomic Precision Diagnostic by Igenomix

Igenomix® WITH SCIENCE ON YOUR SIDE

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABHD12	Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, Cataract	AR	95.77	21 of 21
ADGRV1	Febrile Convulsions, Usher Syndrome, Generalized Epilepsy	AD,AR	97.53	-
ARSG	Usher Syndrome	AR	99.98	2 of 2
CDH23	Deafness, Pituitary Adenoma, Usher Syndrome, Cushing Disease, Prolactinoma	AD,AR	98	400 of 403
CEP250	Cone-Rod Dystrophy, Deafness	AR	99.98	7 of 7
CEP78	Cone-Rod Dystrophy, Deafness, Usher Syndrome	AR	99.44	9 of 10
CIB2	Deafness, Usher Syndrome	AR	99.95	16 of 17
CLRN1	Retinitis Pigmentosa, Usher Syndrome	AD,AR,X,XR,G	99.99	40 of 41
ESPN	Deafness, Usher Syndrome	AR	98.22	22 of 22
HARS1	Charcot-Marie-Tooth Disease, Usher Syndrome	AD,AR	100	-
MYO7A	Deafness, Usher Syndrome	AD,AR	100	579 of 580
PCDH15	Deafness, Usher Syndrome	AR	99.36	152 of 158
PDZD7	Deafness, Usher Syndrome	AR	100	28 of 28
PEX1	Hearing Loss With Enamel Hypoplasia And Nail Defects, Peroxisome Biogenesis Disorder, Zellweger Syndrome, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	97.02	126 of 134
PEX6	Heimler Syndrome, Zellweger Syndrome, Spinocerebellar Ataxia-Blindness-Deafness Syndrome, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AD,AR	99.94	105 of 108
PRPS1	Arts Syndrome, Charcot-Marie-Tooth Disease, Deafness, Phosphoribosylpyrophosphate Synthetase Superactivity, Lethal Ataxia,Intellectual Disability-Limb Spasticity-Retinal Dystrophy-Diabetes Insipidus Syndrome	X,XR,G	100	-
TRNS2	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, Stroke-Like Episodes, Melas, Merrf, Usher Syndrome	MI	-	-
TUBB4B	Leber Congenital Amaurosis With Early-Onset Deafness	AD	100	3 of 3
USH1C	Deafness, Usher Syndrome	AR	99.97	79 of 79
USH1G	Usher Syndrome	AR	100	35 of 35
USH2A	Retinitis Pigmentosa, Usher Syndrome	AR	100	1286 of 1314
WHRN	Deafness, Usher Syndrome	AR	99.94	-

*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. Mathur, P., & Yang, J. (2015). Usher syndrome: Hearing loss, retinal degeneration and associated abnormalities. *Biochimica et biophysica acta*, *1852*(3), 406–420. https://doi.org/10.1016/j.bbadis.2014.11.020
2. Jouret, G., Poirsier, C., Spodenkiewicz, M., Jaquin, C., Gouy, E., Arndt, C., Labrousse, M., Gaillard, D., Doco-Fenzy, M., & Lebre, A. S. (2019). Genetics of Usher Syndrome: New Insights From a Meta-analysis. *Otology & neurotology : official publication of the American Otological Society, American Neurotology Society [and] European Academy of Otology and Neurotology*, *40*(1), 121–129. https://doi.org/10.1097/MAO.0000000000002054
3. Toms, M., Pagarkar, W., & Moosajee, M. (2020). Usher syndrome: clinical features, molecular genetics and advancing therapeutics. *Therapeutic advances in ophthalmology*, *12*, 2515841420952194. https://doi.org/10.1177/2515841420952194
4. Géléoc, G., & El-Amraoui, A. (2020). Disease mechanisms and gene therapy for Usher syndrome. *Hearing research*, *394*, 107932. https://doi.org/10.1016/j.heares.2020.107932
5. Wolfrum, U., & Nagel-Wolfrum, K. (2018). Das Usher-Syndrom, eine Ziliopathie des Menschen [The Usher Syndrome, a Human Ciliopathy]. *Klinische Monatsblatter fur Augenheilkunde*, *235*(3), 273–280. https://doi.org/10.1055/a-0573-9431