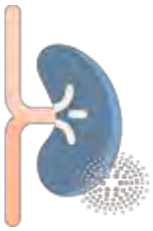
Alport Syndrome

Precision Panel



Overview

Alport Syndrome (AS) is a progressive hereditary renal disease characterized by sensorineural hearing loss, ocular abnormalities and increased risk of chronic kidney failure. It is a genetically and phenotypically heterogeneous disorder of glomerular, cochlear and ocular basement membranes due to a mutation in the genes encoding type IV collagen. Individuals affected by this disease experience progressive loss of kidney function, presenting as blood in the urine (hematuria). The mode of inheritance can be X-linked, autosomal recessive and autosomal dominant.

The Igenomix Alport Syndrome Precision Panel can be used to make a directed and accurate 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 Alport Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of Alport Syndrome presenting with:

- Blood in urine (hematuria)
- Protein in urine (proteinuria)
- Edema
- Hypertension
- Hearing loss
- Ocular manifestations: anterior lenticonus, dot-and-fleck retinopathy, posterior polymorphous corneal dystrophy, temporal macular thinning etc
- Leiomyomatosis

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 symptomatic care, medical care, continuous monitoring of kidney function, and if necessary, renal transplantation.
- Risk assessment 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**
ACSL4	Alport Syndrome-Intellectual Disability-Midface Hypoplasia-Elliptocytosis Syndrome, X-linked Non-Syndromic Intellectual Disability	X,XD,G	99.97	NA of NA
AMMECR1	Alport Syndrome-Intellectual Disability-Midface Hypoplasia-Elliptocytosis Syndrome	X,XR,G	99.81	NA of NA
COL4A3	Autosomal Dominant Alport Syndrome, Autosomal Recessive Alport Syndrome, Benign Familial Hematuria	AD,AR	100	277 of 280
COL4A4	Autosomal Recessive Alport Syndrom, Benign Familial Hematuria	AD,AR	99.95	247 of 251
COL4A5	X-linked Alport Syndrome	X,XD,G	99.88	NA of NA
KCNE5	Alport Syndrome-Intellectual Disability-Midface Hypoplasia-Elliptocytosis Syndrome		99.66	NA of NA

*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. Nozu, K., Nakanishi, K., Abe, Y., Udagawa, T., Okada, S., Okamoto, T., Kaito, H., Kanemoto, K., Kobayashi, A., Tanaka, E., Tanaka, K., Hama, T., Fujimaru, R., Miwa, S., Yamamura, T., Yamamura, N., Horinouchi, T., Minamikawa, S., Nagata, M., & Iijima, K. (2019). A review of clinical characteristics and genetic backgrounds in Alport syndrome. *Clinical and experimental nephrology*, *23*(2), 158–168. https://doi.org/10.1007/s10157-018-1629-4
2. Kashtan C. E. (2021). Alport Syndrome: Achieving Early Diagnosis and Treatment. *American journal of kidney diseases : the official journal of the National Kidney Foundation*, *77*(2), 272–279. https://doi.org/10.1053/j.ajkd.2020.03.026

3. Savige, J., Gregory, M., Gross, O., Kashtan, C., Ding, J., & Flinter, F. (2013). Expert guidelines for the management of Alport syndrome and thin basement membrane nephropathy. *Journal of the American Society of Nephrology : JASN*, *24*(3), 364–375. https://doi.org/10.1681/ASN.2012020148
4. Zhang, Y., & Ding, J. (2017). Renal, auricular, and ocular outcomes of Alport syndrome and their current management. *Pediatric Nephrology*, *33*(8), 1309-1316. doi: 10.1007/s00467-017-3784-3
5. Gross, O. (2008). Understanding renal disorders as systemic diseases: the fascinating world of basement membranes beyond the glomerulus. *Nephrology Dialysis Transplantation*, *23*(6), 1823-1825. doi: 10.1093/ndt/gfn129
6. Savige, J., Ariani, F., Mari, F., Bruttini, M., Renieri, A., Gross, O., Deltas, C., Flinter, F., Ding, J., Gale, D. P., Nagel, M., Yau, M., Shagam, L., Torra, R., Ars, E., Hoefele, J., Garosi, G., & Storey, H. (2019). Expert consensus guidelines for the genetic diagnosis of Alport syndrome. *Pediatric nephrology (Berlin, Germany)*, *34*(7), 1175–1189. https://doi.org/10.1007/s00467-018-3985-4