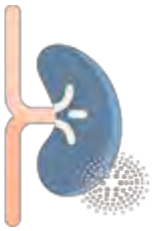
Primary Hyperoxaluria

Precision Panel



Overview

Primary Hyperoxaluria (PH) is a group of inherited metabolic diseases of the liver characterized by increased formation of calcium-oxalate stones in kidneys with the subsequent development of nephrolithiasis and chronic kidney disease. Hyperoxaluria is defined as elevated urinary excretion of oxalate (more than 40mg in 24 hours), a metabolic end product. This elevated excretion can contribute to the formation of kidney stones and other health problems. Mutations in specific liver enzymes involved in the oxalate metabolism make up the etiology of this disease. There are three main types of PH – PH types I, II and III – differentiated by the specific enzyme that is deficient.

The Igenomix Primary Hyperoxaluria Precision Panel can be used to make a directed and accurate diagnosis and aid in the differential diagnosis of recurrent kidney stones 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 Primary Hyperoxaluria Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of Primary Hyperoxaluria presenting with:

- Lower back pain
- Hematuria (blood in urine)
- Pain while urinating
- Inability to urinate
- Increased frequency of urination
- Fever/chills
- Foul smelling urine or cloudy looking urine
- Recurrent kidney stones or urinary tract stones
- Family history of recurrent kidney stones
- Any patient with renal failure of unknown cause

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 medical care to reduce oxalate levels, appropriate hydration and diet changes.
- 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**
AGXT	Primary Hyperoxaluria	AR	99.99	210 of 211
GRHPR	Primary Hyperoxaluria	AR	88	51 of 51
HOGA1	Primary Hyperoxaluria	AR	100	42 of 42

*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. Shah, O., Holmes, R. P., & Assimos, D. G. (n.d.). Management of patients with hyperoxaluria. *Urinary Stone Disease,* 103-119. doi:10.1007/978-1-59259-972-1_7
2. Hoppe, B., Beck, B. B., & Milliner, D. S. (2009). The primary hyperoxalurias. *Kidney International, 75*(12), 1264-1271. doi:10.1038/ki.2009.32
3. Filippova, T. V., Svetlichnaya, D. V., Rudenko, V. I., Alyaev, Y. G., Shumikhina, M. V., Azova, M. M., Subbotina, T. I., Gadzhieva, Z. K., Asanov, A. Y., & Litvinova, M. M. (2019). *Urologiia (Moscow, Russia : 1999)*, (5), 140–143.
4. Milliner, D. (2006). Treatment of the primary hyperoxalurias: A new chapter. *Kidney International, 70*(7), 1198-1200. doi:10.1038/sj.ki.5001821
5. Straub, M., Hautmann, R. E., Hesse, A., & Rinnab, L. (2005). Kalciumoxalatharnsteine und hyperoxalurie. *Der Urologe, 44*(11), 1315-1323. doi:10.1007/s00120-005-0936-z