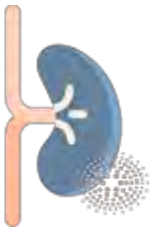
Hemolytic Uremic Syndrome

Precision Panel



Overview

Hemolytic Uremic Syndrome (HUS) is defined by the presence of microangiopathic hemolytic anemia, thrombocytopenia and acute kidney injury. It is one of the main causes of acute kidney injury in children. It is a clinically heterogeneous condition given the different etiologies of HUS that result in differences in presentation, management and outcome. It has been classified into typical HUS resulting from Shiga toxin-producing E.coli infections (STEC) and atypical HUS or non-STEC. HUS can be hereditary due to inborn errors of cobalamin C metabolism, complement gene mutations or acquired by infections, autoantibodies or drug toxicity. It is estimated that approximately 50 percent of non-Shiga-toxin-producing E.coli cases result from mutations in these genes. There is a seasonal pattern of HUS, peaking in the summer and fall. The mode of inheritance follows an autosomal dominant or autosomal recessive pattern.

The Igenomix Hemolytic Uremic 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 Hemolytic Uremic Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis presenting with or without the following manifestations:

- Gastroenteritis: fever, bloody diarrhea
- Irritability
- Lethargy
- Seizures
- Acute renal failure
- Anuria
- Hypertension
- Edema
- Pallor

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 plasma exchange therapy, red blood cell transfusions, appropriate fluid and electrolyte management, provision of adequate nutrition.
- Risk assessment of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASE	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ADAMTS13	Congenital Thrombotic Thrombocytopenic Purpura	AR	99.59	195 of 213
C3	Complement Component 3 Deficiency, Atypical Hemolytic Uremic Syndrome	AD,AR	100	123 of 124
C4BPA	Protein S deficiency, Acute Poststreptococcal Glomerulonephritis	-	98	4 of 4
CD46	Atypical Hemolytic Uremic Syndrome, HELLP Syndrome	AD,AR	100	83 of 84
CD59	Hemolytic Anemia With Or Without Immune-mediated Polyneuropathy	AR	99.99	8 of 8
CFB	Complement Factor B Deficiency, Atypical Hemolytic Uremic Syndrome	AD,AR	100	26 of 26
CFH	Complement Factor H Deficiency, Atypical Hemolytic Uremic Syndrome, HELLP Syndrome	AD,AR,MU,P	99.94	340 of 342
CFHR1	Atypical Hemolytic Uremic Syndrome	AD,AR	88.29	0 of 9
CFHR2	Nonbacterial Thrombotic Endocarditis	-	86.39	4 of 5
CFHR3	Atypical Hemolytic Uremic Syndrome	AD,AR	89.89	0 of 7
CFHR4	Atypical Hemolytic Uremic Syndrome	-	97.3	5 of 6
CFHR5	Cfhr5 Deficiency	AD	100	33 of 34
CFI	Complement Factor I Deficiency, Atypical Hemolytic Uremic Syndrome, HELLP Syndrome	AD,AR	99.93	156 of 158
DGKE	Nephrotic Syndrome	AR	99.67	41 of 42
MMACHC	Methylmalonic Aciduria And Homocystinuria	AR	99.97	105 of 105
PLG	Plasminogen Deficiency	AR	100	79 of 79
THBD	Atypical Hemolytic Uremic Syndrome, Thrombophilia Due To Thrombomodulin Defect	AD	99.91	29 of 30

*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. Noris, M., & Remuzzi, G. (2005). Hemolytic Uremic Syndrome. *Journal Of The American Society Of Nephrology*, *16*(4), 1035-1050. doi: 10.1681/asn.2004100861
2. Fakhouri, F., Zuber, J., Frémeaux-Bacchi, V., & Loirat, C. (2017). Haemolytic uraemic syndrome. *The Lancet*, *390*(10095), 681-696. doi: 10.1016/s0140-6736(17)30062-4
3. Loirat, C., Fakhouri, F., Ariceta, G., Besbas, N., Bitzan, M., & Bjerre, A. et al. (2015). An international consensus approach to the management of atypical hemolytic uremic syndrome in children. *Pediatric Nephrology*, *31*(1), 15-39. doi: 10.1007/s00467-015-3076-8
4. Keir, L. (2015). Shiga Toxin Associated Hemolytic Uremic Syndrome. *Hematology/Oncology Clinics Of North America*, *29*(3), 525-539. doi: 10.1016/j.hoc.2015.01.007
5. Siegler, R., & Oakes, R. (2005). Hemolytic uremic syndrome; pathogenesis, treatment, and outcome. *Current Opinion In Pediatrics*, *17*(2), 200-204. doi: 10.1097/01.mop.0000152997.66070.e9
6. Taylor, C., Machin, S., Wigmore, S., & Goodship, T. (2010). Clinical Practice Guidelines for the management of atypical Haemolytic Uraemic Syndrome in the United Kingdom. *British Journal Of Haematology*, *148*(1), 37-47. doi: 10.1111/j.1365-2141.2009.07916.x
7. Cody, E. M., & Dixon, B. P. (2019). Hemolytic Uremic Syndrome. *Pediatric clinics of North America*, *66*(1), 235–246. https://doi.org/10.1016/j.pcl.2018.09.011