Panel de síndrome urémico hemolítico atípico

Nuestro **panel de síndrome urémico hemolítico atípico** contiene genes para el diagnóstico molecular de este síndrome.

Nº de genes:	25
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	El análisis de CNV incluyó MLPA: CFH, CFHR1, CFHR2, CFHR3, CFHR5

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, hereditary	AR
C3	120700	Macular degeneration, age-related, 9;C3 deficiency;Hemolytic uremic syndrome, atypical, susceptibility to, 5	AR, AD
CD46	120920	Hemolytic uremic syndrome, atypical, susceptibility to, 2	AD, AR
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	AR
CFB	138470	Complement factor B deficiency;Hemolytic uremic syndrome, atypical, susceptibility to, 4;Macular degeneration, age-related, 14, reduced risk of	AR, AD, Digenic dominant
CFD	134350	Complement factor D deficiency	AR
CFH	134370	Basal laminar drusen;Macular degeneration, age-related, 4;Hemolytic uremic syndrome, atypical, susceptibility to, 1;Complement factor H deficiency	AD, AD, AR

CFHR1	134371	Macular degeneration, age-related, reduced risk of;Hemolytic uremic syndrome, atypical, susceptibility to	AD, AD, AR
CFHR2	600889		
CFHR3	605336	Macular degeneration, age-related, reduced risk of;Hemolytic uremic syndrome, atypical, susceptibility to	AD, AD, AR
CFHR5	608593	Nephropathy due to CFHR5 deficiency	AD
CFI	217030	Complement factor I deficiency;Macular degeneration, age-related, 13, susceptibility to;Hemolytic uremic syndrome, atypical, susceptibility to, 3	AR, AD
CR1	120620	[Blood group, Knops system];Malaria, severe, resistance to	
CR2	120650	Immunodeficiency, common variable, 7;Systemic lupus erythematosus, susceptibility to, 9	AR
DGKE	601440	Nephrotic syndrome, type 7;Hemolytic uremic syndrome, atypical, susceptibility to, 7	AR
F12	610619	Factor XII deficiency;Angioedema, hereditary, type III	AR, AD
G6PD	305900	Resistance to malaria due to G6PD deficiency;Hemolytic anemia, G6PD deficient (favism)	XLD
INF2	610982	Glomerulosclerosis, focal segmental, 5;Charcot-Marie-Tooth disease, dominant intermediate E	AD
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMUT	609058	Methylmalonic aciduria, mut(0) type	AR
PIGA	311770	Paroxysmal nocturnal hemoglobinuria, somatic;Multiple congenital anomalies-hypotonia-seizures syndrome 2	XLR
PLG	173350	Angioedema, hereditary, 4;Plasminogen deficiency, type I;Dysplasminogenemia	AD, AR
PRDX1	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic	AR
THBD	188040	Thrombophilia due to thrombomodulin defect;Hemolytic uremic syndrome, atypical, susceptibility to, 6	AD
VWF	613160	von Willebrand disease, types 2A, 2B, 2M, and 2N;von Willebrand disease, type 1;von Willebrand disease, type 3	AD, AR, AD, AR

CFHR1	134371	Macular degeneration, age-related, reduced risk of;Hemolytic uremic syndrome, atypical, susceptibility to	AD, AD, AR
CFHR2	600889		
CFHR3	605336	Macular degeneration, age-related, reduced risk of;Hemolytic uremic syndrome, atypical, susceptibility to	AD, AD, AR
CFHR5	608593	Nephropathy due to CFHR5 deficiency	AD
CFH	134370	Basal laminar drusen;Macular degeneration, age-related, 4;Hemolytic uremic syndrome, atypical, susceptibility to, 1;Complement factor H deficiency	AD, AD, AR