Test for atypical Hemolytic Uremic Syndrome (aHUS)
What is atypical Hemolytic Uremic Syndrome (aHUS)?

It is an ultra-rare genetic thrombotic microangiopathy mainly affecting the kidneys. The majority of aHUS patients present recurrent disease and more than 50% develops chronic kidney disease.

The aHUS is fundamentally associated with mutations and SNPs (single nucleotide polymorphisms) in genes encoding proteins of the complement system such as CFH, MCP (CD46), CFI, CFB, C3, among others.

What is included in the test of aHUS performed by SECUGEN?

**GENETIC STUDY**
- Next Generation sequencing panel for CFH, CFI, MCP (CD46), CFB, C3, THBD, DGKE, CFP, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5 and ADAMTS13 genes.
- Analysis of rearrangements in CFH/CFHR1-5 region.
- Genotyping of risk/protection haplotypes for CFH and MCP (CD46) genes.
- Comparison with proprietary database.

**BIOCHEMISTRY AND FUNCTIONAL STUDY**
- ELISA quantification. Nephelometry of factor H, factor I and factor B levels.
- Flow cytometry quantification of MCP (CD46). DAF.
- Factor H functional analysis.
- Search for anti-FH autoantibodies.

Additionally we offer Sanger sequencing for CFH, CFI, MCP (CD46), CFB, C3, THBD, DGKE, CFP and ADAMTS13 genes.

**Advantages of this study**
- Provides prognostic value of aHUS evolution.
- Assists therapeutic and management discussions.
- Helps selection of live related donors.
- Eases genetic counseling and risk assessment in relatives.

Secugen gets advice and support from the Work Group in Complement and Renal Pathology, an international reference in complement’s studies, coordinated by Drs. Rodríguez de Córdoba, López-Trascasa and Sánchez-Corral Gómez.

**References**