Abstract Submission No. : 9105
May 28(Sat), 10:40-12:40 Genetic Disease

Complementopathy in Atypical HUS/C3 Glomerulopathy

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Atypical hemolytic uremic syndrome (aHUS) is a rare disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, and acute renal failure. The pathology of aHUS is thrombotic microangiopathy (TMA), in which there is extensive thrombosis in arterioles and capillaries; In addition, C3 glomerulopathy (C3G) is a rare set of glomerulonephritis with 2 patterns: C3 glomerulonephritis (C3GN) and dense deposit disease. The dysregulation of the alternative pathway of the complement activation system, either from acquired or genetic cause, is the underlying mechanism of both diseases. Recent studies of the genetic background of them have revealed significant genetic heterogeneity in several genes involved in complement regulation, including complement factor H, CFH-relate protein, complement factor I, complement factor B, complement component 3 and CD46 genes in the alternative complement pathway.

Although clinical studies have provided a better understanding of disease pathogenesis, genetic factors are still largely unknown, and it is difficult to identify genetic predispositions in various clinical conditions. In addition, given the prognosis of the diseases remains poor, further research is needed to improve diagnosis and treatment protocols based on genetic predisposition and to develop new therapeutic agents.