#1952 - MHY9 gene polymorphisms in a patient with macro-platelet thrombocytopenia and renal failure

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Body	Combination of thrombocytopenia with renal disease happens in diverse clinical situations. Genetic variations in MYH9 gene can cause MYH9-related thrombocytopenia (MYH9-RD). A case diagnosed as idiopathic thrombocytopenic purpura and his renal study was compatible with focal segmental glomerulosclerosis (FSGS). We studied the whole exons of MYH9 gene by next-generation sequencing for this patient. Single nucleotide polymorphisms (SNPs) in the introns 13 (rs3752462) and 14 (rs2413396), and a mutation in exon 26 of MYH9 gene were observed. Our result supported the possibility of non-coding SNPs involvement in the pathogenicity of the MYH9-RD. It is important to consider the possibility of macro-platelet-thrombocytopenia in patients with thrombocytopenia and renal involvement otherwise unnecessary immunosuppressive therapy creates deleterious side effects without bringing any benefit.
References	Balduini CL, Pecci A, Savoia A: Recent advances in the understanding and management of MYH9-related inherited thrombocytopenias. Br J Haematol 2011;154:161-174.
Generate date	03 November 2018 14:52