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A rare case of association between Fabry's nephropathy and membranous glomerulonephritis: New perspectives on pathophysiology and follow-up of Fabry's disease

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Fabry disease (FD) is a rare X-linked disorder resulting from the deficiency of alpha-galactosidaseA enzyme. Microalbuminuria is the initial manifestation of renal involvement, progressing to end-stage renal disease. From one case, we followed the patient's response to enzyme replacement therapy (ERT) and the evolution of its manifestations. A 61 years old male was referred to nephrologist to investigate generalized edema and massive proteinuria. He referred a previous diagnosis of cardiomyopathy and heart failure treatment. Physical examination revealed widespread edema. Complementary tests showed nephrotic proteinuria, hypoalbuminemia and dyslipidemia. Renal biopsy revealed membranous glomerulonephritis (MN) and FD association. Anti-phospholipase-A2-Receptor autoantibodies were positive, revealing the unprecedented association between idiopathic MN and Fabry nephropathy, reinforces the hypothesis that Fabry's nephropathy may modify podocyte antigens, leading to idiopathic MN. Others FD manifestations were found: cornea verticillata, hypertrophic cardiomyopathy and supratentorial microangiopathy. The α-Gal activity was reduced, associated with lyso-Gb3 accumulation. Genetic analysis identified an unreported hemizygous mutation in exon 7 of the GLA gene. The patient experienced decreased edema and clinical stabilization with the institution of fortnightly ERT with agalsidase alfa, with complementary exams showing preservation of renal function with reduction in proteinuria and increased serum albumin. Family screening identified six close relatives with FD on oligosymptomatic stage. This study recognized an unknown association between MN and FD and an unreported genetic mutation. It's also serving as the basis for the development of a database that aims to allow the follow-up of these patients, making possible the analysis of clinical data and of its evolution.

Biography

Daniel Santos Rocha Sobral Filho is a Medical Student at Federal University of Piauí, Teresina - Piauí – Brazil and has Scholarship of the Program of Scientific Initiation of the Federal University of Piauí. He participates in researches in nephrology, focusing on genetic nephropathies.

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