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Immunoglobulin A Nephropathy in a patient with Neurofibromatosis Type 1

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Case Study:

Neurofibromatosis type 1 is an autosomal dominant neuro-cutaneous disorder involving skin, bones, and nervous system. The most common kidney involvement is renal artery stenosis, and glomerulonephritis is extremely rare. In this case, we report a case of NF-1 with Immunoglobulin A Nephropathy (IgAN).

A 51-year old Korean man previously diagnosed with neurofibromatosis type 1 presented with persistent proteinuria and hematuria on his routine medial checkup. He had no history of hypertension or diabetes, and alcohol or smoking history was denied by the patient. On the contrast enhanced CT, both kidney was intact in size, and there was no evidence of renal artery stenosis. On the day of kidney biopsy, laboratory data showed serum creatinine 1.1 mg/dL, urine protein/creatinine ratio 1.3 g/g, and urine RBC 10~15>/HPF. Kidney biopsy confirmed IgA nephropathy, grade III by Lee's glomerular grading system.

Three cases of NF-1 and IgAN had been reported in Japan, and a few case of membranous nephropathy and focal segmental glomerulosclerosis had been reported in patients with NF-1. Genetic link between NF-1 and IgAN or other type of glomerulonephritis is not certain however, activation of mTOR pathway due to the inadequate suppression of p21-ras protein resulted from the deficient neurofibromin possibly interprets the link.