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LMX1B mutation cause hereditary focal segmental glomerulosclerosis without extrarenal manifestation

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Case Study:

Background
Mutations in *LMX1B* cause nail-patella syndrome (NPS), an autosomal dominant disorder, characterized by nail dysplasia, skeletal abnormality, and nephropathy. Recent genetic studies have shown that some specific missense mutations in *LMX1B* cause isolated nephropathy without overt orthopedic symptoms. We present a case of focal segmental glomerulosclerosis (FSGS) without orthopedic symptoms in a patient with *LMX1B* mutation.

Case presentation
A 52-year-old woman was admitted to our hospital for an investigation of the cause of her proteinuria and microscopic hematuria. On physical examination, she was normotensive and had a pretibial pitting edema. She has a normal renal function and the results of other serological tests for screening of vasculitis and infection were negative. The spot urine protein-to-creatinine ratio (PCR) was 4.6 g/g. She had a family history of renal disease: father was on hemodialysis for end stage renal disease (ESRD) of unknown cause; younger brother was being treated for FSGS; and daughter underwent a kidney transplant for ESRD by FSGS. She was diagnosed with FSGS by renal biopsy, and a genetic analysis showed a mutation in *LMX1B* gene. Considering the possibility of NPS, we re-checked physical examination of the musculoskeletal system. However, we could not find specific orthopedic manifestation. Angiotensin II receptor blocker and SGLT2 inhibitor were prescribed for her. After 6 months, her renal function remained within the normal range and spot urine PCR decreased to 2.0 g/g.

Conclusions
We have described a case of *LMX1B* mutation associated FSGS without extrarenal involvement. Isolated FSGS can be caused by mutations in genes that are also involved in syndromic forms of the disease. Therefore, it is necessary to consider including these genes in all diagnostic approaches of FSGS.