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A novel missense mutation identified in a Korean family with X-linked Alport syndrome (AS) using next generation sequencing (NGS)

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Case Study: We herein report a novel mutation in a Korean family with an X-linked Alport syndrome (AS) mutation in COL4A5. A 15-year old Korean male presented to nephrology clinic for evaluation of proteinuria and hematuria. The patient had a family history for kidney problem in his maternal grandfather and his mother has hearing loss. His sister also had proteinuria and hematuria. Physical examination revealed a blood pressure of 110/60mmHg with no peripheral edema. Laboratory evaluation showed a serum creatinine of 0.89mg/dL, 24-hour urine protein of 3,819mg and serum albumin of 3.8 g/dL. A renal biopsy confirmed the diagnosis of AS. We performed targeted next generation sequencing (NGS) to identify mutations associated with AS. The most likely disease-causing variants were identified and confirmed by Sanger sequencing. This is the first reported case of a patient with AS in missense mutation (c.2332G> C, p.Gly778Arg).