# Nephrotic Syndrome Developing in a Girl with Classic 21-Hydroxylase Deficiency – First Report

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## *Objectives:*

Background: Nephrotic syndrome is the most common cause of kidney disease in children, but its pathogenesis remains unclear. Nephrotic syndrome in patients with congenital adrenal hyperplasia has not been reported.

Objectives: To present a case of nephrotic syndrome occurring in a chinese girl with classic 21-hydroxylase deficiency.

## *Methods:*

Case presentation.

## *Results:*

A 38-month-old female child was admitted with eyelid edema. She was the first child (birth weight, 3.0 kg, full term) of non-consanguineous parents of Chinese Han ethnicit. She had been diagnosed with congenital adrenal hyperplasia due to classic 21-hydroxylase deficiency, salt-wasting type in the neonatal period. Mutation analysis demonstrated that the patient was a compound heterozygote with mutations of paternal c.293-13C>G and maternal c.60G>A(p.Trp20X) in *CYP 21A2* gene. With good adherence to steroid replacement treatment with hydrocortisone and fludrocortisone, she was evaluated to be in 'Good Control'based on clinical criteria including signs of androgen excess, growth velocity and bone age increment. Laboratory examination showed that ALB was 17.4 g/L. Urinalysis showed the presence of proteinuria (+++) without hematuria. In addition, the 24-h urine protein was 1.52 g/day (95 mg/kg per day), serum creatinine was normal, triglyceride was 3.5 mmol/L, cholesterol was 9.6 mmol/L. Hepatitis B/C serology were negative. Complement C3 and C4 level were normal. The patient was diagnosed with idiopathic nephrotic syndrome, and treated with oral prednisone instead of hydrocortisone. Remission of proteinuria was attained after 5 days. Regular hydrocortisone and fludrocortisones for 21OHD were given after 5-month prednisone treatment. To this day, the patient has been followed up for 18 months with remisson of proteinuria.

## *Conclusions:*

We first report a steroid-responsive idiopathic nephrotic syndrome occurring in 38-month girl with classic 21-hydroxylase deficiency. The relationship between nephrotic syndrome and congenital adrenal hyperplasia could contribute to the pathogenesis of nephrotic syndrome.

## *References:*

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