Nephrotic Syndrome Developed in a Girl with Lipoid Adrenal Hyperplasia due to StAR gene mutation – First Report

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Introduction

Lipoid congenital adrenal hyperplasia is the most severe form of congenital adrenal hyperplasia. It is most commonly caused by mutations in steroidogenic acute regulatory protein.
And the incidence of idiopathic nephrotic syndrome (NS) is 1.5~16.9 per 100,000 children.
NS is characterized by the triad of proteinuria, hypoalbuminemia, and edema. There can be an antecedent infection, typically of the upper respiratory tract. Moreover, NS can be accompanied with several complications such as infection, thromboembolism and hypovolemic crisis.
Here, we report a nephrotic syndrome with hypotension occurring in 10 years old girl with primary adrenal insufficiency.

Case

- **10Y old Girl**

- **P/I**

She was admitted with generalized edema and drowsy mentality.
She got fever and mild extremities edema 2 days before admission, and revealed generalized edema and drowsy mentality on admission.

- **P/Hx**

- She had been diagnosed and taking medicines (hydrocortisone, fludrocortisone) due to primary adrenal insufficiency since neonatal period.
- She had admission history for idiopathic nephrotic syndrome two year ago.
- Recently, she get the genetic test for primary adrenal insufficiency, which showed StAR gene homozygous mutation (c.772C>T,p.Q258X), diagnosed as lipoid adrenal congenital hyperplasia.

- **V/S**

BP 90/40mHg, HR 120~130/min, RR 25~30/min, BT 39℃.

- **G/A**

Acutely ill app(+)
Puffy face(+), Extremity fitting edema(+)

- **Course in hospital**

A 10-years old girl was admitted in intensive care unit with generalized edema and mental change. She was the first baby (birth weight, 3.0kg, full term) of non-consanguineous parents.
The patient had a decreased blood pressure and tachycardia.
Table 1. showed the laboratory investigation results upon admission.
It was assumed that adrenal crisis and hypovolemic crisis due to idiopathic nephrotic syndrome affected the patient hypotension and drowsy mentality simultaneously. The patient was initially treated with high dose intravenous hydrocortisone (100mg a day). Her hypotension and drowsy mentality recovered on the second day of hospitalization, and then, intravenous hydrocortisone was changed into oral prednisolone. Remission of proteinuria was achieved after 8th day of hospitalization.
The patient has been followed up for about 1 year with remission, until the present.

Laboratory investigations	
CBC	15,770/μL/ 12.7 g/dL/ 341K/ μL
Serum Na/K	129 mg/dL/ 5.3 mg/dL
AST/ALT	34/20 U/L
BUN/Cr	18.0 mg/dL / 0.8 mg/dL
Protein/Albumin	3.7 mg/dL/1.59 mg/dL
Total cholesterol	280 mg/dL
Glucose	50 mg/dL
Renin activity	19.5 ng/ml/hr (n: 0.6-4.18)
Aldosterone	3.51 ng/dL (n: 11.58~41.98)
Urinalysis	
Protein(++++) , OB(-), RBC 0-2/HPF , WBC 0-2/HPF	
Spot Pr/Cr 42.5	

Table.1. The patient's laboratory investigations on admission

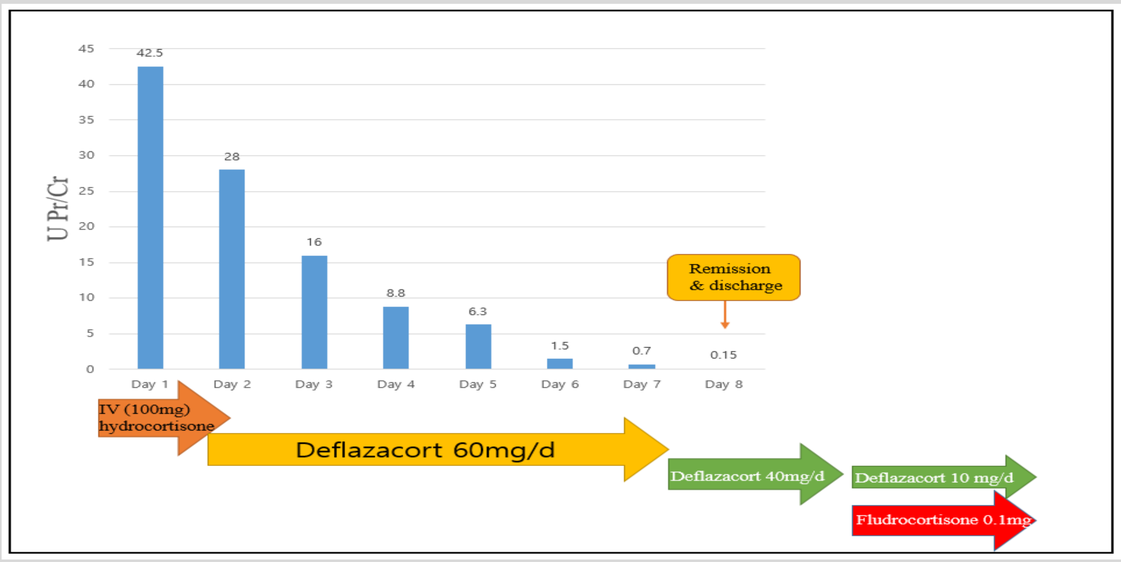

Table.2. Hospital course of the case. The patient was initially treated with high dose hydrocortisone, and then which was changed to oral prednisolone.

Proband	c.772C>T (homozygous)
Father	c.772C>T (heterozygous)
Mother	c.772C>T (heterozygous)

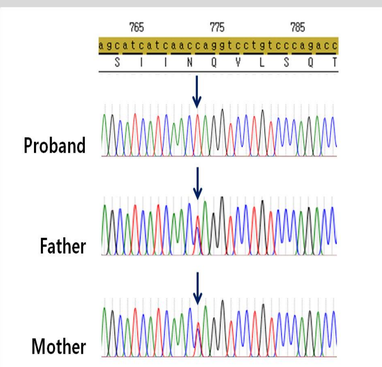

Fig.1. The patient was diagnosed with the homozygous variant c.C772T on StAR gene developing stop codon at codon 258 in exon 7

Conclusion

It is difficult to know whether two diseases have occurred accidentally or not. However, it was thought that both NS and adrenal crisis could contribute to hypotension and drowsy mentality of the patient. We first report a idiopathic nephrotic syndrome presenting hypotension and drowsy mentality, needed with high dose intravenous hydrocortisone developed in a girl with lipoid congenital adrenal hyperplasia.