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CASE REPORT

HDR syndrome presented with nephrotic syndrome in a Chinese boy: A case report

Li-Juan Ma, Wu Yang, Hong-Wen Zhang

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Abstract

BACKGROUND

HDR syndrome is a rare genetic disease caused by variants in the *GATA3* gene and is phenotypically defined by the triad of hypoparathyroidism (H), deafness (D), and renal disease (R). Renal disorders of HDR are mainly developmental abnormalities, although renal functional abnormalities can also be observed. Nephrotic syndrome or nephrotic-level proteinuria is rare in HDR syndrome. Here, we report a Chinese infant with HDR syndrome who presented with early-onset nephrotic syndrome. We suggest that variants in the *GATA3* gene might be associated with nephrotic syndrome.

CASE SUMMARY

A 9-month-old boy was hospitalized with a complaint of diarrhea. Proteinuria was detected in the patient by routine testing for 3 days. No edema, oliguria, fever or abnormal urine color were observed. Routine urinary tests at a local hospital revealed proteinuria (protein 3 +) and microscopic hematuria (red blood cells 5-10/HP). The patient was born by cesarean delivery due to placental abruption at 35 weeks + 4 days of gestation. Intrauterine growth retardation was detected beginning at 6 months of gestation. His birth weight was 1.47 kg (< P3th), length was 39 cm (< P3th), and head circumference was 28 cm (< P3th). His motor developmental milestones were obviously delayed. Clinical data were analyzed, and genetic analysis for hereditary nephrotic syndrome was performed by next-generation sequencing. The clinical data showed that the boy exhibited growth retardation, early-onset nephrotic syndrome, microscopic hematuria, sensorineural deafness, T-cell immunodeficiency and congenital heart disease. Genetic tests revealed that the boy carried a *de novo* hemizygous variant, c.704C>T

(p.Pro235 Leu), in exon 3 of the *GATA3* gene.

CONCLUSION
We report an infant with HDR syndrome who presented with early-onset nephrotic syndrome in China. We suggest that variants in the *GATA3* gene might be associated with infant-onset nephrotic syndrome.

Key Words: HDR syndrome; Sensorineural deafness; Nephrotic syndrome; China; Case report

Core Tip: HDR syndrome is a rare genetic disease caused by variants in the *GATA3* gene and is phenotypically defined by the triad of hypoparathyroidism (H), deafness (D), and renal disease (R). Patients with HDR syndrome may exhibit the full phenotypic triad or only a subset. Renal disorders of HDR are mainly developmental abnormalities, although renal functional abnormalities can also be observed. Nephrotic syndrome or nephrotic-level proteinuria is rare in HDR syndrome. Here, we report a Chinese infant with HDR syndrome who present with early-onset nephrotic syndrome.

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INTRODUCTION

HDR syndrome (OMIM 146255) is a rare autosomal dominant genetic disease caused by variants in the *GATA3* gene located on chromosome 10p14. GATA-3 plays an essential role in the embryonic development of the parathyroids, inner ear and kidneys. HDR syndrome is phenotypically defined by the triad of hypoparathyroidism (H), deafness (D), and renal disease (R)[1,2]. HDR syndrome has been reported previously in China[3,4] and other Asian countries[2,5,6]. Here, we report a Chinese boy with HDR syndrome who presented with early-onset nephrotic syndrome.

CASE PRESENTATION

Chief complaints

A 9-month-old boy was hospitalized with a complaint of diarrhea.

History of present illness

Proteinuria was detected in the patient by routine testing for 3 days. The parents denied any occurrence of edema, oliguria, fever or abnormal urine color. Routine urinary tests at a local hospital revealed protein 3 + and red blood cells 5-10/HP.

History of past illness

The boy was born to nonconsanguineous healthy parents. He was born by cesarean delivery due to placental abruption at 35 weeks + 4 days of gestation. Intrauterine growth retardation was detected beginning at 6 months of gestation. His birth weight was 1.47 kg (< P3th), length was 39 cm (< P3th), and head circumference was 28 cm (< P3th). His motor developmental milestones were obviously delayed. He was able to raise his head at 4 months. He was unable to turn over, crawl or sit at 9 months.

Personal and family history

There was no abnormal past medical or family history, and no family history of maternal renal disease.

Physical examination

At admission, the patient's weight was 7.1 kg (< P3th), height was 66 cm (< P3th), and head circumference was 36 cm (< P3th). He was able to make sounds, look and hear with audiovisual stimulation. No obviously abnormal dysmorphic features were revealed in physical examination. No positive signs were found on examination of his lungs, heart and abdomen. No other neurodevelopmental or ophthalmologic deficits were observed.

Laboratory examinations

The results of routine laboratory tests, including routine blood test, electrolytes, liver and kidney function, thyroid

hormone, growth hormone and insulin-like growth factor 1, were all normal. All autoantibodies were negative. The levels of C3 and C4 were normal while the levels of immunoglobulin G (IgG) and IgA were decreased. T/B lymphocyte subsets showed some abnormalities. The total parathyroid hormone level was normal. Routine urinary tests displayed protein 3 + and red blood cells 5-10/HP. The urinary microalbumin concentration ranged from 4384-5981 mg/L, and the spot urinary protein-to-creatinine ratio ranged from 2.96-5.87 (Table 1). The results of blood and urine metabolism analyses were all normal. According to the Gesell development schedules, the estimated total DQ was 72, the adaptive behavior DQ was 78, the large motor behavior DQ was 65, the fine motor behavior DQ was 66, the language behavior DQ was 75, and the personal social behavior DQ was 76. Pure tone audiometry indicated sensorineural deafness. His karyotype was 46, XY.

Imaging examinations

Ultrasonography revealed that the renal body was normal (6.2-6.6 cm in length and 2.4-3.1 cm in width), and the border of the cortex and medulla was clear. Echocardiography revealed an atrial septal defect (5.0 mm). Electroencephalography and cranial magnetic resonance imaging results were normal.

Genetic analysis

Genetic analysis was performed using next-generation sequencing at the genetics laboratories of the MyGenostics biotechnology company in China, using "the inherited renal diseases panel", which covers genes strongly correlated with this disorder. The results showed that the boy carried a hemizygous variant, c.704C>T (p.Pro235 Leu), in exon 3 of the *GATA3* gene, and his parents were both wild type (Figure 1). The variant was not found in the Human Gene Mutation Database or the SNP databases including ALFA, ExAC, GnomAD and TOPMED. PolyPhen-2, Variant Taster and GERP++ analysis showed that the variant was pathogenic, while SIFT analysis indicated that the variant might be benign (ACMG guideline: PS2 + PM2_Supporting).

FINAL DIAGNOSIS

Due to the presence of growth retardation, early onset nephrotic syndrome, microscopic hematuria, sensorineural deafness, abnormal immune functions, congenital heart disease and c.704C>T (p.Pro235 Leu) in the *GATA3* gene, HDR syndrome was diagnosed.

TREATMENT

Following the diagnosis of HDR syndrome caused by *GATA3* variant, there was no specific treatments.

OUTCOME AND FOLLOW-UP

The patient did not receive any specific treatment after the diagnosis of HDR syndrome caused by a *GATA3* variant. Over a follow-up of 3 months, there was no obvious change in proteinuria (urinary microalbumin: 4695-5739 mg/L, urine protein/creatinine: 3.05-6.21), microscopic hematuria (urine red blood cells: 5-10/HP) or growth retardation.

DISCUSSION

Renal disorders of HDR mainly involve developmental abnormalities. However, renal functional abnormalities can also be observed. Here, we report the case of a Chinese infant with HDR syndrome and renal disease, including early-onset nephrotic syndrome and microscopic hematuria.

To date, approximately 180 HDR syndrome cases have been reported worldwide. Over 90% of patients present with hypoparathyroidism and sensorineural deafness, and more than 80% of patients exhibit urinary tract and renal abnormalities. Patients with HDR syndrome may exhibit the full phenotypic triad or only a subset. Our patient presented with growth retardation, early-onset nephrotic syndrome, microscopic hematuria, sensori-neural deafness, T-cell immunodeficiency and congenital heart disease (atrial septal defect). Genetic analysis revealed a *de novo* heterozygous variant, c.704C>T (p.Pro235 Leu), in exon 3 of the *GATA3* gene. Based on his clinical manifestations and genetic results, HDR syndrome was diagnosed. Interestingly, our patient presented without hypoparathyroidism. In rare cases, further manifestations have been reported in HDR syndrome, including female genital tract malformation, retinitis pigmentosa, growth retardation, pyloric stenosis, neurological abnormalities, T-cell immunodeficiency[6-9] and congenital heart disease[5,9]. Our patient also exhibited growth retardation, T-cell immunodeficiency and congenital heart disease.

Renal disorders of HDR syndrome include both developmental abnormalities (such as renal hypoplasia, dysplasia, aplasia, cystic kidney disease, pelvicalyceal deformity, and vesicoureteral reflux) and functional abnormalities (such as proteinuria, hematuria, glomerulonephritis, proximal or distal renal tubular acidosis, and nephrocalcinosis)[1-3,10]. Nephrotic syndrome and nephrotic-level proteinuria are rare in HDR syndrome patients. Chenouard *et al*[11] reported a child with HDR syndrome with nephrotic syndrome as a novel finding. The first renal biopsy at 3 years revealed tubuloi-

Table 1 Laboratory findings of the patient during hospitalization

Items	Results	Normal range
Total serum protein	39.40 g/L	55-75
Serum albumin	24.40 g/L	39-54
Total cholesterol	5.89 mmol/L	< 6.22
Triglyceride	2.08 mmol/L	< 1.7
Serum creatinine	18.40 μmol/L	13-33
Blood urea nitrogen	3.54 mmol/L	1.1-5.9
Serum calcium	2.32 mmol/L	2.1-2.8
IgG	1.07 g/L	7-16
IgA	0.28 g/L	0.7-5
IgM	0.60 g/L	0.4-2.8
C_3	1.12 g/L	0.78-2.1
C_4	0.22 g/L	0.17-0.48
Total parathyroid hormone	22.22 ng/L	12-88
25-OH vitamin D	62.15 μg/L	30-100
Total T cell	62.44%	56-71
CD4+ T cell	44.23%	25-39
CD8+ T cell	16.71%	20-34
CD4/CD8	2.65	0.76-1.61
B cell	31.15%	13-25
NK cell	6.44%	8-20
Urinary microalbumin	4384-5981 mg/L	0-15
Urine protein/creatinine	2.96-5.87	< 0.2
Urine red blood cell	5-10/HP	< 3

IgG: Immunoglobulin G; IgA: Immunoglobulin A; IgM: Immunoglobulin M; NK: Natural killer.

nterstitial nephritis without dysplasia or glomerular injury, while the second renal biopsy at 4 years revealed segmental, diffuse, proliferative glomerulonephritis and persistent nephrocalcinosis. Maleki *et al*[12] reported a 58-year-old male patient with HDR syndrome with chronic kidney disease and nephrotic-level proteinuria (2.99 g/24 hours). He had a family history of kidney disease in his siblings, whose renal biopsies revealed focal segmental glomerulosclerosis. The patients described by Barakat *et al*[13] presented with steroid-resistant nephrotic syndrome, renal histology revealed fetal-like glomeruli and thickened glomerular basement membranes. The renal disorder in our boy fulfilled the criteria for early-onset nephrotic syndrome, with a serum albumin concentration of 24.40 g/L and a urinary protein/creatinine ratio of 2.96-10.86 g/g. Unfortunately, the renal pathology of our patient was unclear because his parents did not consent to a kidney biopsy.

Overall, 10% of HDR patients progress to end stage renal disease[14]. The age at which renal dysfunction occurs in HDR syndrome patients is variable. In the report of Chenouard *et al*[11], renal failure was detected with a serum creatinine 107 μmol/L at 3 years of age, but without obvious progression in 25 years (serum creatinine: 121 μmol/L). In the patient reported by Maleki *et al*[12], chronic kidney disease occurred at 56 years of age with a serum creatinine concentration of 221 μmol/L. Horta *et al*[15] reported that a 51-year-old male patient with HDR syndrome who presented with mild kidney disease (serum creatinine 186 μmol/L), and his father died at the age of 69 years of chronic kidney disease. According to the report by Joseph *et al*[16], a 47-year-old mother was found to have impaired renal function (serum creatinine: 246 μmol/L) and chronic parenchymal changes on renal scan. According to the report by Barakat *et al*[13], four siblings died from end-stage renal disease between the ages of 3 and 8. Our patient's renal function is currently normal, and follow-up is needed as the patient ages.

The mechanism of GATA-3 pathogenesis in the renal involvement of HDR syndrome remains unclear, and to date, no potential target of this transcription factor has been identified. However, *GATA3* is expressed in mouse kidney mesangial cells, and is markedly increased in rodent models of mesangial proliferative glomerulonephritis, suggesting that *GATA3* plays a critical role in normal glomerular development and might be a useful nuclear marker of human mesangial cells [17]. In the last 20 years, 133 *GATA3* variations have been reported; patients have shown great clinical variability, and the

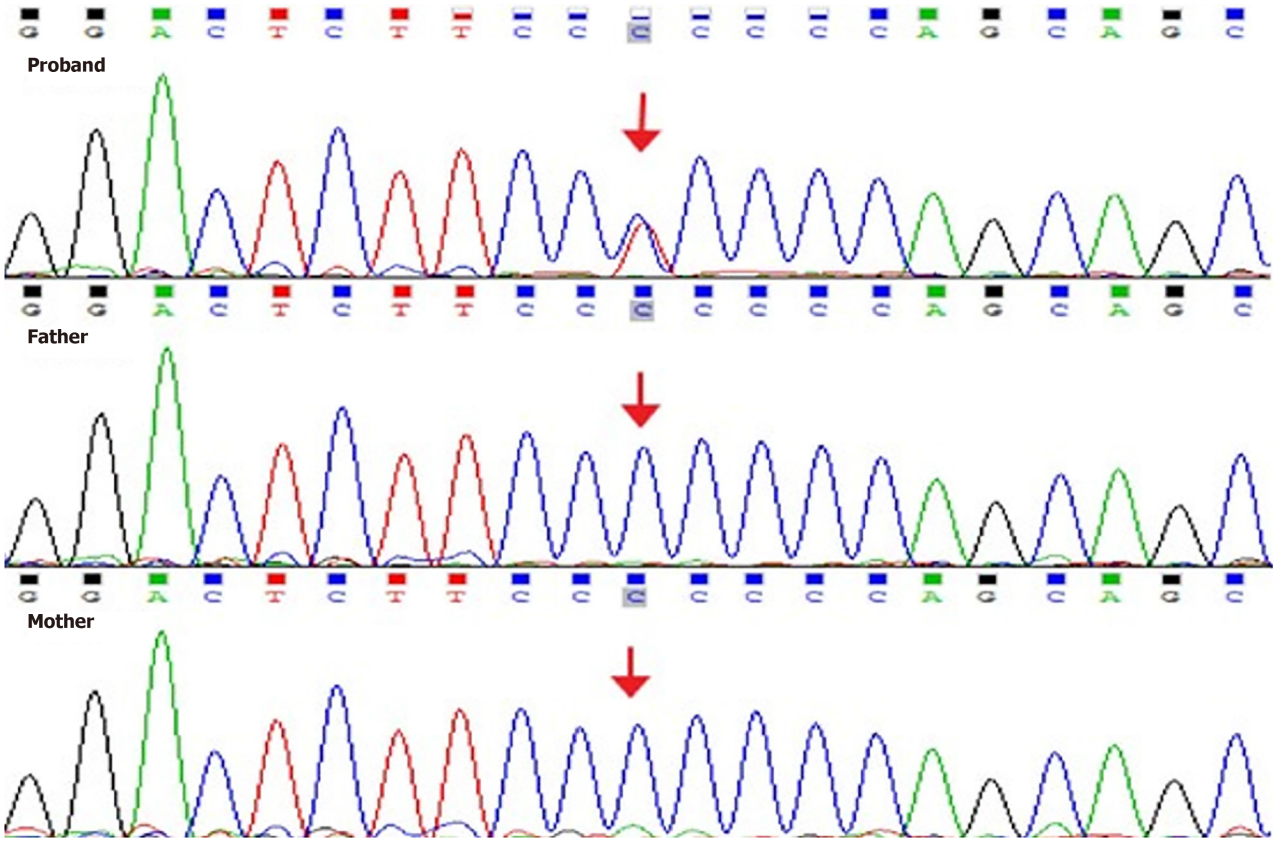

Figure 1 Genetic analysis of the *GATA3* gene. The results revealed that the boy carried a *de novo* heterozygous variant, c.704C>T (p.Pro235 Leu), in exon 3 of the *GATA3* gene.

penetrance of each HDR defect increases with age[18]. However, no clear genotype and phenotype correlation has been determined[19].

CONCLUSION

Here, we report a Chinese infant with HDR syndrome who presented with early-onset nephrotic syndrome, microscopic hematuria, sensorineural deafness, growth retardation, abnormal immune function and congenital heart disease. We suggest that variants in the *GATA3* gene might be associated with infant-onset nephrotic syndrome, which extends the spectrum of phenotypes of *GATA3* disorders. Screening for *GATA3* variations is therefore relevant for patients with either two or three of the phenotypic manifestations of HDR syndrome. Further studies of *GATA3* are needed to improve our knowledge of the involvement and phenotype of this transcription factor in human development, particularly in the kidneys.

FOOTNOTES

Author contributions: Ma LJ collected the data and wrote the manuscript; Yang W and Zhang HW performed the diagnosis and treatment; Zhang HW guided the diagnosis and treatment and revised the manuscript; Ma LJ and Yang W contributed equally to this work and should be considered co-first authors.

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