INTRODUCTION

Nephronophthisis (NPH) is an autosomal recessive disease characterized by chronic tubulointerstitial nephritis that progresses to end-stage renal disease in children and adolescents. The juvenile form manifests by urine concentration defect during the first decade of life and 20% of patients have additional extrarenal manifestations. Currently, the definitive diagnosis is made by molecular genetic approach. All NPH gene products are expressed in the primary cilia and centrosomes, therefore NPH is considered to be a ciliopathy.

CASE REPORT

♀, 9 year-old

Present history
Uncontrolled vomiting

Physical examination
Moderate dehydration
NORMAL blood pressure
High urinary output

Past history
Hyperopia
Astigmatism
Hypotonia
Intellectual disability
Cerebellar dysplasia

Detailed history
Several years ago
Polyuria
Polydipsia
Enuresis

Urinary concentration defect + CKD

CILIOPATHY ?

Neurologic disorder
Ophthalmologic disorder

Conservative therapy

Renal Ultrasound
Bilateral echogenic kidneys
Loss of corticomedullary differentiation

Genetic Test
Bialelic deletion on NPHP1

Renal biopsy – not performed

Analysis

AKI ?

Haemoglobin
10,1 g/dL
Urea
142 mg/dL
Creatinine
2,1 mg/dL
(GFR 26,55 mL/min/1.73m²)
Sodium
139 mEq/L
Urine Specific Gravity
1005

Evolution

MAINTAINED Kidney injury

Urea
101 mg/dL
Creatinine
1,85 mg/dL
(GFR 30,14 mL/min/1.73m²)

Urinary concentration defect

CONCLUSION

This case highlights the diagnostic challenge of NPH because of its unspecific clinical presentation and significant overlap with other ciliopathies. A high index of suspicion is necessary, particularly when multisystemic involvement is present. A low urine specific gravity in a dehydrated patient with a severe renal insufficiency points out to a possible underlying tubular defect. In our case, these features combined with the other extrarenal manifestation suggested the diagnosis.

References:

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