Urinary stone disease is not so rare in children. The aim of this study was to assess the demographic, clinical and biological characteristics, as well as outcome, of urinary stone disease among Croatian children. We reviewed medical records of 76 children from various parts of Croatia who were diagnosed with urinary stone disease from 2002–2011. The average age (mean) was 9 yr 7 mo (toddlers 7.89%) with approximately equal gender distribution (male 53.95% vs female 46.05%). Family affection was identified in 27 (35.53%) children with the predominance of female transmission. The most stones were made of Ca oxalate dihydrate and monohydrate (75%), Hyperoxaluria were detected in 47.57%, mild hyperoxaluria in (13.16%), hypoxaturia in 1.31% and 38.16% remained of idiopathic origin. Urine saturation (EQUIL 2) were above the limits in 47 (61.84%) children, urine volume less than average in 12 (15.79%). For most of the children we recommended increased fluid intake and balanced food nutrition, citrate were administered in 20 (26.32%), thiazides in 10 (13.15%) and aldosetone in 1 (1.31%). Spontaneous evacuation were noticed in 51.32%, surgical (operation and endoscopic removal) in 11.84%, ESWL in 11 0.84%, spontaneous resolution (cetraxone) in 1 (1.31%) and in 15.16% the stone was not removed from urinary tract. The study gave insight to etiology of urinary stone disease in Croatian children. Main pathological factors were hypercalciuria, mild hyperoxaluria and increased urine saturation. Spontaneous evacuation of stones were notified for most of children.

1209 VESICOURETERAL REFLUX AND URODYNAMIC Dysfunction

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The concept of vesicoureteral reflux (VUR) as a consequence of a congenital anomaly of the vesicoureteral junction have undergone changes owing to the finding that such children may have a lower urinary tract dysfunction, which produces a high intravesical pressure and consequently a predisposition for VUR. We investigated relationship of VUR and urodynamics. The urodynamics was investigated by pressure-flow-EMG study in 132 children with primary VUR and 162 refluxing units. Only 33 (25.0%) patients had normal urodynamic finding. The most frequent pathological finding was overactive bladder (OAB), found in 59 (44.7%) children, followed by dysfunctional voiding (DV) in 25 (18.9%) children. The children with VUR grades I and II had higher percentage of pathological urodynamic findings than children with VUR grades III and IV. OAB was more frequent in children under 5 years of age, with unilateral and lower grades VUR. It was found equally in children with and without uroinfections. DV was more frequent in children older than 5 years, with bilateral VUR, higher grades VUR and uroinfections. The results of our study show that the children with VUR have high incidence of urodynamic disorders and indicate the possible role of urodynamic dysfunction in the pathogenesis of VUR, especially the mild ones. They also indicate the need for incorporating urodynamic investigation in the evaluation of children with VUR.

1210 URINARY STONE DISEASE IN CHILDREN- A SINGLE CROATIAN CENTER EXPERIENCE

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In children, idiopathic nephrotic syndrome is primarily treated using corticosteroids. When remission is not achieved, the administration of potent immunosuppressant therapy becomes imperative. Cyclosporine A (CyA) is reportedly associated with a higher incidence of remission in comparison with other immunosuppressive agents. The aim of our study is to evaluate the efficiency of cyclosporin A (CyA) therapy in 11 children treated with resistant nephrotic syndrome.

Methods Eleven children enrolled in this study were all hospitalized with resistant nephrotic syndrome, aged 1 to 11 years (average 5.8 yrs) and included 7 males and 4 females. CyA was given to each patient with dosage of 5 mg/kg/day during the corticosteroid was diminished. The renal biopsy was performed in all patients before the administration of CyA.

Results Eleven children with resistant nephrotic syndrome of different pathological types were treated with CyA, including 3 cases of minimal change nephrotic syndrome (MCNS), 2 cases of mesangiproliferative glomerulonephritis (MsPGN), 1 case of extra membranous glomerulonephritis (EMGN) and 5 cases of focal segmental glomerular sclerosis (FSGS).

Three patients got complete remission, seven patients developed chronic renal insufficiency and one had no change after four month treatment with CyA. The overall response rate was 27%. Patients with different renal pathological types showed different responses. The FSGS cases showed the lowest rate.

Conclusion CyA has limited efficiency in patients with steroid-resistant nephrotic syndrome. CyA should be used cautiously because of the potential for CyA nephrotoxicity.

1212 GITELMAN SYNDROME IN A SPANISH GYPSY PAEDIATRIC PATIENT MUTATION INTRON 9 +1G-T

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Introduction

Gitelman syndrome is an inherited tubular disorder characterized by metabolic alkalosis, hypokalemia and hypomagnesemia of renal origin and hypocalciuria. The majority of patients with Gitelman syndrome carry inactivating mutations in the SLC12A3 gene encoding the sodium-chloride cotransporter located in the distal convoluted tubule. The purpose of this report is to describe a new mutation of the SLC12A3 gene in a gypsy boy, mutation of ancient origin that would be specific in this ethnic group and spread throughout Europe.

Case Report

A 5 years old male children of Roma origin (Gypsy) was referred to our hospital because asthenia, muscle weakness and hypokalemia. Both parents are healthy, non consanguineous with normal serum potassium. There were no other family members affected. Relevant biochemical data at diagnosis was: Serum: pH 7.52, bicarbonate 31mmol/L, potassium 2 mEq/L, sodium 136 mEq/L, chloride 97 mEq/L, magnesium 1.6mg/dl, creatinine 0.4 mg/dl, calcium 9 mg/dl. Plasma Renin Activity 13.3 ng/ml/h, Aldosterone 138 pg/ml.

Urinary potassium 51 mEq/L, calcium/creatinine ratio 0.12; Potassium fractional excretion 20.4%, magnesium fractional excretion 5.9%. Renal ultrasonography and blood pressure was normal.

Genetic study was performed: the patient was homozygous for splice site mutation guanidine to thymine in the first position of intron 9 of SLC12A3 gene (intron 9+1G>T).

Conclusion

This finding will facilitate the identification of the genetic defect in further cases of Gitelman syndrome among the gypsy population. This patients exhibit muscle sympotms and asthenia althoug the disease is not particulary severe in this ethnic group.

Background and Aims

We report a 4-year-old boy with a history from the first months of life characterized by failure-to-thrive, hyporexia, polydipsia and polyuria, and delayed motor function. In recent months she was unable to ambulate due to progressive muscle weakness, especially of the lower limbs. Physical findings included a severe weight and height delay, signs of severe malnutrition and rickets, and tachypnea. Laboratory findings were consistent with distal renal tubular acidosis (metabolic acidosis with normal anion gap, severe hypokalemia, hypophosphatemia, mild hypocalcemia, and hypouricaemia, with urinary pH > 6 and positive urinary gap). Also, bilateral renal nephrocalcinosis was found. Alkali replacement was able to correct the electrolyte abnormalities and promote catch-up.

Comments

dRTA must be suspected in the presence of the clinical and laboratory findinds reported.