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Recurrent carpopedal spasms and numbness over limb extremities in an 11 year old

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Introduction

Gitelman's syndrome (GS) is an autosomal recessive primary renal tubular disorder which commonly presents in adolescent and adult age group.1 Metabolic alkalosis, hypokalemia, hypomagnesemia and hypocalciuria are the cardinal biochemical abnormalities of GS.2 Common clinical features of GS are easy fatigability, muscle cramps and carpopedal spams.1 Here, we present a girl with Gitelman's syndrome who presents with carpopedal spasms.

Case report

An 11-year-old previously healthy girl presented with recurrent carpopedal spasms and numbness over limb extremities for 2-year duration. These episodes were associated with febrile illnesses and symptoms spontaneously settled when she became afebrile. In addition, there was a history of polyuria and polydipsia for the same duration.

She was the first child born to non-consanguineous healthy parents at term with a birth weight of 3.8kg. There was no history suggestive of antenatal polyhydramniosis. Since early childhood her growth had been suboptimal for which she was not investigated. She had no history of urinary tract infections, recurrent vomiting or history of diuretic ingestion. There was no family history of renal disorders or history of muscle weakness or electrolyte disturbance. Her development had been age appropriate and the immunization was up to date.

On examination, her height was 135 cm (10th Centile), Weight was 23.5 kg (<5th Centile) and her Body Mass Index (BMI) was 12.8 kg/m2 (<5th centile). She has no dysmorphic features and her hydration status was good. There were no abnormalities in her cardiovascular system examination including blood pressure. Rest of her system examination was normal.

Her basic haematological parameters were normal. Biochemical investigations revealed hypokalemic, hypochloremic metabolic alkalosis with hypomagnesemia. Urinary electrolytes showed elevated urinary sodium, potassium and chlorides. She had low urine calcium: creatinine ratio suggestive of hypocalciuria. Her ultrasound scan of the kidneys revealed no abnormality. Her serum renin and aldosterone levels were normal. Electrocardiogram didn't show any features of hypokalaemia such as ST depression or U waves.

Investigation	Value	Reference range
Serum Sodium	139 mmol/L	(135-145)
Serum Pottasium	2.8mmol/L	(3.5-4.5)
Serum Chloride	88 mmol/L	(90 - 110)
PH	7.48	(7.35-7.45)
HCO_3	30 mmol/L	(22-26)
PCO_2	48 mmHg	(35-45)
Serum Calcium	2.3 mmol/L	(2.1- 2.57)
Serum Magnesium	1.6 mmol/L	(1.7 – 2.7)
Urine Sodium	244 mmol/L	(20 - 110)
Urine Potassium	15.7 mmol/L	(10 -60)
Urine Chloride	153 mmol/L	(55 -125)
Urine Osmolality	215 mOsm/ kg	(50 - 1050)
Urine Calcium: Creatinine ratio	0.92 mmol/l	(2.5 -7.5)
Serum Creatinine	63 µmol/l	(60 - 110)
Blood Urea	32 mg / dl	(19 - 44)
Serum Renin	33.2 µU/ml	(4.4 - 46)
Serum Aldosterone	26.3 ng/ml	(4 - 31)

The diagnosis of Gitelman's syndrome was made in the presence of characteristic biochemical abnormalities, normal blood pressure and the clinical presentation. Genetic mutation analysis was not done due to limited resources. She was commenced on Magnesium oral potassium and Spironolactone. Following treatment her symptoms were subsided with the improvement of biochemical abnormalities.

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Discussion

GS was first described by Hillel J. Gitelman in 1966 and the disease was named after him.3The disorder is commonly due to the genetic mutation of SLC12A3 gene or rarely due to CLCNKB gene.1 The genetic mutations impair the action of the sodium chloride co-transporter at the distal convoluted tubule of the nephron resulting excess urinary loss of sodium, potassium and chloride.4 The hallmark biochemical abnormalities in the GS are metabolic alkalosis, hypokalemia, hypomagnesemia and hypocalciuria.2 In the past, GS was considered as a variant of Bartter Syndrome (BS), which shares common features of GS such as hypokalemia and hypochloremic metabolic alkalosis. However, in BS hypomagnesemia and hypocalciuria are not seen and the pathophysiology of the condition due to a defect in the Sodium, Potassium, Chloride co transporter at the thick ascending limb of the loop of Henle.[5]

Though, GS is an inherited condition, patients with the specific genetic mutation remain asymptomatic until late childhood or early adolescence.[1] Like in this child, the most common presenting feature is carpopedal spasms during episodes of vomiting and febrile illnesses.[4] Moreover, patients with GS present with paraesthesia, muscle cramps and easy fatigability. In this child, majority of these clinical features were present at the time of presentation. In addition, there are reported cases of GS presents with cardiac arrest.[6]
The diagnosis of GS is mainly based on the clinical presentation and the characteristic biochemical abnormalities.[1] However, it is important to exclude conditions which can mimic the condition such as diuretic abuse or recurrent vomiting.[2] In this child, there was no history suggestive of diuretic abuse or recurrent vomiting. The confirmatory test of the condition is DNA mutational analysis, which was not done in this case due to non availability of the test in government sector.

There is no cure for the condition and the aim is to correct electrolyte abnormalities.[7] High salt diet and oral potassium and Magnesium supplements are the existing therapeutic interventions. In some cases, potassium sparing diuretics such as Spironolactone or Amiloride can be used. In this child, Spironolactone was used as a potassium sparing agent.[7] Prostaglandin inhibitors such as Indomethacin or Ibuprofen and Angiotensin Converting Enzyme Inhibitors such as Captopril can be used in advanced cases of GS.

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