1213 A BOY WITH HYPERKALEMIA AND HYPERTENSION WITHOUT FAMILY HISTORY: STILL PSEUDOHYPOALDESTERONISM (GORDON SYNDROME)?

Introduction Gitelman syndrome is an inherited tubular disorder characterized by metabolic alkalosis, hypokalemia and hypomagnesemia of renal origin and hypocaliuria. 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 presence 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 symptoms and asthenia although the disease is not particularly severe in this ethnic group.

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1214 SEVERE RICKETS AND HYPOKALEMIC FLACCID PARALYSIS DUE TO DISTAL RENAL TUBULAR ACIDOSIS (dRTA) IN A 4-YEAR-OLD GIRL

Introduction Distal renal tubular acidosis (dRTA) is a tubulopathy characterized by metabolic acidosis with normal anion gap secondary to a defective secretion of H+ ions by the collecting tubule. This anomaly leads to an inability to acidify the urine during systemic acidemia. There are more than 90 different mutations, with an autosomal recessive or dominant pattern of transmission. Clinical features usually appear from two years of age as vomiting, dehydration and failure to thrive, although the first signs may be present from the first weeks of life. In recessive forms can be associated with sensorineural deafness. The prognosis is favorable if alkali replacement is performed properly. However, urolithiasis, nephrocalcinosis and chronic renal failure may appear if the diagnosis is delayed or the treatment is inadequate.

Case Report We report a 4-year-old Moroccan girl with a history from the first months of life characterized by failure-to-thrive, hypoxemia, polypenia and polyuric, 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 hypouricemia, with urinary pH > 6 and positive urinary gap). Also, bilateral renal nephrocalcinosis was found. Alkali replacement was able to correct the electrolytic abnormalities and promote catch-up.

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

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1215 REFERENCE VALUES OF SERUM CYSTATIN C IN VERY LOW BIRTH WEIGHT PREMATURE INFANTS

Background and Aims To determine reference values for cystatin C (CysC) and its correlation with creatinine (Cr), gestational age, birth weight and maternal Cr status in very low birth weight (VLBW) preterm infants.

Aims The study included 113 VLBW premature infants (< 1500 g) of 53 gestational week.

Results The mean level of CysC was 1.77±0.38 mg/L on day 1 and 1.61±0.37 mg/L on day 3, and the decrease was statistically significant. There was a significant correlation only between maternal Cr and first-day Cr values and negative correlations between Cr and gestational age and birth weight on third day. Creatinine was not correlated with CysC both on day 1 (r = -0.077, p=0.417) and day 3 (r=0.132, p=0.164).

Conclusion CysC offer an important advantage in the measurement of renal functions independent from gestational age, birth weight and maternal Cr status in VLBW preterm infants.

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1216 INTRAUTERINE GROWTH RESTRICTION AND DEVELOPMENTAL PROGRAMMING OF RENAL DISEASE

Abstracts