¹B Orive, ²J Montiano, ³C Salado, ³A Torrents. ¹*Pediatric Nephrology*; ²*Pediatrics, Hospital Universitario de Álava, Vitoria*; ³*Genetics, Reference Laboratory, Barcelona, Spain*

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 SLC 12A3 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 guanine 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.

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

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¹RA Hollander, ¹D Trouet, ²G Mortier. ¹*Pediatric Nephrology*; ²*Medical Genetics, Antwerp University Hospital, Edegem (Antwerp), Belgium*

Background and Aims We report a 6-year-old boy presenting with macroscopic hematuria, hyperkalemia and hyperphosphatemia.

Results A 6-year-old boy presented to our outpatient clinic with hematuria. No clinical abnormalities were found, besides borderline hypertension of 120/60mmHg (P95 adjusted for height and age 100/57). Laboratory testing revealed a serum potassium of 8.6 mmol/l and a phosphate of 6.2mg/dl. During admission hyperkalemia appeared refractory to standard treatment with furosemide, salbutamol and alkalisation. Prompt reduction of hyperkalemia was observed after a trial with hydrochlorothiazide, suggesting the diagnosis of pseudohypoaldosteronism. Further testing revealed a high aldosterone of 537 pg/ml. Familial history was negative but still a suspicion of pseudohypoaldosteronism remained, so genetic testing was performed. A de novo splice site mutation was found in the CUL3-gene (c.1377+1G>A) leading to a skipping of exon 9.

Conclusions We present an atypical presentation of Gordon syndrome (familial hyperkalemic hypertension) due to a de novo splice mutation. The clinician should be aware of this possibility when facing a child with high serum potassium.

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

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JD Martinez-Pajares, O Diaz-Morales, J Trigo-Moreno, JC Ramos-Diaz, M Miranda-Valdivieso, E Fernandez-Gomez. *UGC Pediatria, Hospital Comarcal de Antequera, Antequera, Spain*

Background 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 50 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, 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 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.

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

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

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¹G Demirel, ¹H Celik, ¹FE Canpolat, ²O Erdeve, ³Z Biyikli, ¹U Dilmel. ¹*Neonatology*; ²*Zekai Tahir Burak Maternity and Teaching Hospital*; ³*Biostatistics, Ankara University*; ⁴*Pediatrics, Yildirim Beyazit University, Faculty of Medicine, Ankara, Turkey*

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 ≤32 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). The reference values of Cys C at birth are [median (3p-97p)]: 1.8 mg/dl (0.8-2.2) for 24-26 weeks, 1.8 mg/dl (1.51-3.19) for 26-28 weeks, 1.8 mg/dl (0.65-2.48) for 28-30 weeks and 1.79 mg/dl (0.68-2.31) for 30-32 weeks and at 3rd days are 1.5 mg/dl (0.54-2.0) for 24-26 weeks, 1.61 mg/dl (1.1-3.4) for 26-28 weeks, 1.7 mg/dl (0.56-2.3) for 28-30 weeks and 1.6 mg/dl (0.92-2.21) for 30-32 weeks.

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.

1216 INTRAUTERINE GROWTH RESTRICTION AND DEVELOPMENTAL PROGRAMMING OF RENAL DISEASE

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