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 symptoms and asthenia although the disease is not particularly severe in this ethnic group.

### Abstracts

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

DOI: 10.1136/archdischild-2012-302724.1213

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

**Background and Aims** We report a 6-year-old boy presenting with macroscopic hematuria, hypokalemia and hyperparasitaemia.

**Results** A 6-year-old boy presented to our outpatient clinic with hematuria. No clinical abnormalities were found, besides borderline hypertension of 120/60mmHg (95% 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, 0.5mg/kg/day hydrochlorothiazide, 1mg/kg/day salbutamol and alkalinisation. 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 site 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**

DOI: 10.1136/archdischild-2012-302724.1214

1G Demirel, 1IH Celik, 1FE Canpolat, 1D Erdeve, 2Z Biyikli, 1U Dilmen. 'Neonatology; 2Zekai Tahir Burak Maternity and Teaching Hospital; 1Biostatistics, Ankara University; 1Pediatrics, 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.

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

**Aims** 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 CysC at birth are [median (3p-97p)]: 1.8 mg/dl (0.8–2.2) for 24–26 weeks, 1.8 mg/dl (1.5–3.19) 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.
Background and Aims  Intrauterine growth restriction (IUGR) complicated by umbilical artery vasospasms may have adverse effects on integrity and function of vascular endothelium and has been associated to atherosclerosis and to glomerulosclerosis risk in adulthood. Aortic intima media thickening, a preclinical sign of atherosclerosis, has been documented from intrauterine life, whereas the first signs of glomerulosclerosis have not been determined. So the aim of this study was to compare albuminuria/creatininuria ratio (ACR) in IUGR and appropriate for gestational age (AGA) neonates.

Methods  A prospective cohort study has been performed on 25 IUGR consecutive fetuses evaluated at Department of Woman and Child Health of Padua University between December 2009 and December 2010. They were considered IUGR if the estimated fetal weight (EFW) was below 10th percentile and the umbilical artery pulsatility index (PI) > 2 standard deviations. Each IUGR newborn was matched with 2 controls AGA neonates, of the same gestational age ± 1 week. They were considered AGA if the EFW was between 10th and 90th percentile. A urine sample was collected at 24–72 hours after birth for ACR determination.

Results  Among the 25 IUGR fetuses enrolled, 2 were excluded because I had trisomy 21 and 1 renal agenesis. The remaining 23 were matched with 46 AGA newborns. ACR was significantly higher in IUGR compared to AGA newborns: median (IQR) 183.0 (113.6–264.7) vs 122.8 (72.5–191.9); p=0.04.

Conclusions  IUGR is associated with significantly greater albuminuria at birth. This may be an early marker of glomerulosclerosis, which leads to renal disease in adulthood.

1219 DIFFERENCES IN MORTALITY/MORBIDITY WITH A COMPLETE COURSE OF ANTENATAL STERIODS COMPARED TO AN INCOMPLETE/NO COURSE IN EXTREMELY PREMATURE NEONATES

doi:10.1136/archdischild-2012-302724.1219

1D Wong, 1ME Abdel-Latif, 1AL Kent. 1Australian National University, Medical School; 2Dept of Neonatology, Canberra Hospital, Canberra, ACT, Australia

Background Antenatal steroids have been shown to reduce mortality and morbidity in neonates born less than 29 weeks gestation. Counselling of parents regarding outcomes is generally based on data that includes all neonates whether they have received a complete course of antenatal steroids or not. In the acute setting where delivery is imminent more accurate estimation of outcomes for parents is required.

Aims To determine the differences in survival, short and long term morbidity for those neonates receiving no antenatal steroids or an incomplete course of steroids in comparison to those receiving a complete course.


Results 2549 neonates were included in the study with 319 (12.8%) not given any ante-natal steroids. Hospital mortality was significantly worse without steroids (30% versus 20%; p=0.001). Those with no steroid coverage were more likely to have NEC (11% vs. 7%; p=0.018) and Grade 3 or 4 IVH (19% vs 12%; p=0.001). In a multivariate model, factors predictive of mortality included: lack of antenatal steroids, male gender, smaller gestation and hypertensive disease of pregnancy.

Long-term data was available for 1473 survivors. There was no difference in long term neurological outcome between those not receiving steroids and those receiving any steroids.