

**Manchester Medical Society  
MANCHESTER PAEDIATRIC CLUB**

**Abstracts for the Scientific Meeting**

**MANCHESTER ACADEMIC HEALTH  
SCIENCES CENTRE  
Research (5<sup>th</sup> Floor)  
St Mary's Hospital  
Oxford Road  
Manchester, M13 9WL**

**Thursday 6<sup>th</sup> October 2011  
9.30-11.30 am  
(Coffee & Registration from 9.00 am)**

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**Dr Saiel Saleh**

## THINK OF APPARENT MINERALOCORTICOID EXCESS IN CHILDREN WITH HYPERTENSION, HYPOKALAEMIA AND RENAL CYSTS

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A 4 year old Asian girl of consanguineous parents presented with severe hypertension (BP 160/120mmHg). Previously, under Cardiological care for pulmonary hypertension, she was noted to have left ventricular hypertrophy. Born at 29 weeks, she was small for gestational age (820g, 2<sup>nd</sup> centile). There was a long history of failure to thrive, polydipsia and polyuria. Ultrasonography identified medullary nephrocalcinosis with two cysts 5-6mm across in the right kidney and a single cyst in the left. Serum potassium was low (3.1 mmol/L) and bicarbonate elevated (31 mmol/L) and there was hypercalciuria (calcium/creatinine 2.5; normal <0.7). Urinary catecholamine screen was normal.

Plasma aldosterone concentration (<5 5pmol/L; NR: 300-1830) and renin activity (<0.2 ng/ml/hr; NR: 1.7-3.9) were low. Urinary steroid profiling identified an increased tetrahydrocortisol/tetrahydrocortisone ratio, characteristic of apparent mineralocorticoid excess (AME). The diagnosis was confirmed by identifying a homozygous pathological mutations in *HSD11B2* which codes for 11 $\beta$ HSD2, an enzyme in the placenta and kidney which inactivates cortisol by conversion to cortisone. Amiloride therapy ameliorated polyuria/polydipsia, corrected electrolyte imbalances, and improved her hypertension. Parental renal ultrasonography was normal, making autosomal dominant polycystic kidney disease unlikely, and we failed to find *HNF1B* mutation, a not uncommon cause of renal cysts in children.

Ours is the 4<sup>th</sup> report of renal cysts in AME syndrome. Previously, cystogenesis has been ascribed to chronic hypokalaemia and renal cysts can indeed feature in other hypokalaemic disorders. Fetuses with *HSD11B2* mutations will be exposed to glucocorticoid excess and, experimentally, glucocorticoids cause cysts in maturing kidneys. We postulate that in AME syndrome, an abnormal fetal milieu primes renal cystogenesis which is accentuated postnatally by hypokalaemia. AME syndrome also illustrates that not all cystic renal disease is caused by ciliopathy gene mutations but that environmental factors are also important.

**HYPOGLYCAEMIA IN CHILDREN WITH ACUTE  
GASTROENTERITIS- WHERE DOES IT GO WRONG?  
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**Introduction**

Numbers of children with hypoglycaemia complicate acute gastroenteritis (AGE) in East Lancashire Hospital NHS Trust increased rapidly from 2008 to 2010. NICE produced a guideline on management of diarrhoea and vomiting in children less than five in 2009. We hypothesized that NICE guideline gave ambiguous sugar advice in their fluid recommendation and causing medical staffs and parents to give inappropriate fluids to children with AGE.

**Method**

57 parents and 105 medical staffs from Emergency Department (ED), Paediatric Department (PD) and General Practice were surveyed. Percentage of medical staffs and parents who gave NICE compliance and sugar-contained fluids were calculated. We then compare answers from parents who never seek help (A), received advice (B) and never received advice (C) from doctors and medical staffs of different specialities. Data was analysed using StatsDirect.

**Result**

4.41% children with AGE had hypoglycaemia in 2010 compared to 1.26% in 2008. There was difference in practice between different specialities. 64.1%, 19.0% and 35.0% medical staffs in ED, PD and GP gave NICE compliant advice and 71.8%, 97.6% and 95.0% gave sugar-contained fluids. 66.7%, 69.2%, and 57.9% of parent group A, B and C complied with NICE guideline and 55.6%, 53.8% and 68.4% gave sugar-contained fluids.

**Conclusion**

We concluded that NICE guideline gave ambiguous recommendation regarding sugar in the fluids for children with AGE. This ambiguity has caused inappropriate administration of no sugar-contained fluids to children with AGE and thus causing the increase in numbers of hypoglycaemia complicates AGE.

# GENETIC INVESTIGATION OF CHILDREN PRESENTING WITH VELOPHARYNGEAL INSUFFICIENCY (VPI)

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Indistinct or nasal speech is a common reason for children to present to a speech and language therapist. Around 68% of these children are diagnosed with velopharyngeal insufficiency (VPI). This is an abnormality of palate function, where the soft palate is unable to adequately close off the back of the pharynx during speech. Many children with VPI have other problems such as learning difficulties or behavioural problems and may therefore be referred for genetic assessment. This study investigated 42 children who were diagnosed with VPI plus additional features and referred for genetic assessment. Each child had a full medical and family history, a clinical examination and appropriate investigation including routine karyotype and/or microarray analysis. Of the 42 children, 1 had an abnormal karyotype and 16 had an abnormality detected on microarray analysis. In total, 17 patients (40.5%) had a pathogenic chromosomal abnormality. The commonest underlying diagnoses were 22q11.2 deletion (23.5%), 16p11.2 deletion (23.5%) and 22p11.2 duplication (11.8%) with the remainder having other sporadic cytogenetic abnormalities. Family studies on 2 other patients suggested that they had single gene disorders. Further patients had distinctive but undiagnosed multiple anomaly syndromes. For patients receiving a diagnosis, the impact of this on further management was assessed. These results suggest that underlying genetic disorders are common in children with VPI and additional features. Dissemination of this information to the speech and language team is recommended to raise awareness and encourage appropriate referral to the genetic clinic.

PSYCHOLOGICAL SUPPORT MEASURABLY IMPROVES QoL IN CF PATIENTS. Andy Peeling 1 Meadowfield Preston Lancs PR2 9RE

**Objective:** To investigate whether quality of life is better in patients who receive psychological support (thus proving its efficacy) after a diagnosis of cystic fibrosis; to exclude the issue of community deprivation as a direct influence; and to provide proof-of-concept for larger-scale studies with similar randomisation constraints. **Cohort:** 24 patients with formally diagnosed CF ( $n_{\text{treatment}}=12$ ,  $n_{\text{control}}=12$ ). **Method:** The CFQ-UK (Cystic Fibrosis Questionnaire) was administered to either patient, parent, or both. Where possible, the SDQ (Strengths and Difficulties Questionnaire) was administered to parents of the patients. **Results:** 34 CFQ questionnaires were completed. The mean indices of deprivation were worse in the treatment-naive group, but correlation between these and each questionnaire measure was not significantly different from zero. Patients in the treatment group scored better on average than the treatment-naive group in all domains except body image. Mann-Whitney U tests for the mean SDQ scores showed a median improvement of 20% for hyperactivity, 10% for emotional and peer-related problems and 10% for pro-sociality but 0% improvement for conduct problems (95.7% CI). The CFQ-UK demonstrated no median improvement in the domains of weight, eating and digestion but significant improvements in treatment behaviours (22.2%), general health (11.2%), respiratory health (8.35%), physical health (8.3%) and emotional wellbeing (7.5%) (95.5% CI). **Conclusion:** Deprivation was not significantly causative in our sample. Patients with CF who receive ongoing psychological support in East Lancashire enjoy a better quality of life than those who do not. Therapy sessions may need to take a different approach to address the issue of eating behaviours. A larger-scale study needs to take place to reproduce this effect.

**CONGENITAL HEART DISEASE AND ASSOCIATED RISK FACTOR: A COHORT STUDY OF THE BLACKBURN WITH DARWEN POPULATION (2006-2010). Roslan, AF, Sibley A, Khan N**

University of Manchester School of Medicine, UK. Paediatric department, East Lancashire Hospital Trust, UK.

**Introduction:** A perceived large number of children are identified to have congenital heart disease (CHD) within the Blackburn with Darwen area. Currently children with CHD in the area receive coordinated medical care but not all families receive genetic counselling. The survey aims to explore the incidence and patterns of congenital heart disease (CHD) within the area. The survey also aims to explore associated risk factors for congenital heart disease including ethnicity, and consanguinity.

**Method:** Patients diagnosed with CHD born from 2006-June 2011 were identified through the East Lancashire Hospitals Trust's and Alder Hey Childrens' Foundation's Trust record. Only patients with a Blackburn and Darwen postcode were included in the study. Characteristics of their medical diagnoses and demographic details were recorded anonymously from medical records. Results were analysed by using Statsdirect.

**Result:** There were 90 patients diagnosed with CHD from 2006-Jun 2011. 37.8% were asian-Pakistani, 57.8% were Caucasian, 3.3% Indian and 1.1% other. The incidence of CHD was 31.9 per 10,000 livebirths in 2009 and 48 per 10,000 livebirths in 2010. 78.46% of cases were complex and 21.54% were simple. CHD is strongly associated with the asian-Pakistani ethnic ( $p < 0.0001$ ). There is strong association between complex CHD and consanguineous marriage ( $p = 0.0237$ ). There were 4.9% stillbirth and 16.05% mortality rate.

**Conclusion:** There is a high incidence of CHD within the Blackburn and Darwen population in 2009 and 2010. CHD is more common in people of asian-Pakistani origin. There is substantial evidence that complex CHD is associated with parents of consanguineous marriage. This may have implications for local provision of medical and genetics services.

**PILOT STUDY TO EXPLORE AND DESCRIBE THE PERCEIVED NEEDS AND CONCERNS OF CHILDREN WITH COMPLEX NEEDS AS THEY GRADUATE FROM SPECIAL SCHOOLS AND TRANSITION TO THE ADULT SERVICES.**

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**INTRODUCTION:** This paper describes the characteristics of the population of children graduating from special schools in East Lancashire and their transition to adult services. This survey is a pilot for a more extensive study into the perceived needs and service provision for this population.

**METHODS:** Case notes of 26 graduates from Newfield Special School were used for descriptive data and analysed using SPSS. Qualitative data regarding perceived needs, and experiences of planning and service provision were collected from 8 parents over the phone using an anonymous questionnaire. Additional data was obtained from 6 healthcare professionals involved in transition.

**RESULTS:** Amongst 26 children, 40% had no specific diagnosis, however 96.2% had learning difficulties. Transition plans were present in 73% of cases. Most parents expressed satisfaction with the transition process although lack of information and difficulty when navigating different agencies involved was often quoted. Healthcare professionals pointed to the lack of resources and guidance to individuals as the main concerns in transition.

**CONCLUSION:** Personalisation and patient-centred care are the main aspects perceived as needing improvement. Despite increase in awareness and improvements to transition planning, communication and coordination gaps between agencies were highlighted. A larger survey could define standards for identification, documentation and communication of individual needs and the provision for categories of need which would then be amenable to audit.

**RAPID TAPERING OF CORTICOSTEROIDS POST PAEDIATRIC RENAL TRANSPLANTATION:  
ASSESSMENT OF GROWTH AT 1 YEAR. AN INTERIM ANALYSIS.**

- **Authors**

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- **Abstract**

Corticosteroids constitute a vital component of the immunosuppression regime after renal transplantation in the paediatric patient. However, prolonged steroid exposure can contribute to growth suppression. The recent TWIST study showed that early steroid withdrawal significantly aided growth at 6 months without increasing graft rejection or loss. Here, we present 1 year follow up data from patients at the Royal Manchester Children's Hospital.

Children received tacrolimus, MMF, 2 doses of basiliximab and steroids until day 4 (n=7) (SCS), or tacrolimus, azathioprine, and standard course steroids (n=10) (LCS). Mean changes in height SDS (standard deviation score) for SCS was 0.4, and 0.23 for LCS. Frequency of biopsy-proven acute rejection was 28.6% in SCS and 30.0% in LCS. Patient and graft survival were identical, with similar renal function at 1 year.

The initial analysis suggests the growth advantages of early steroid withdrawal shown in the TWIST study at 6 months are still present at 1 year, without impacting on frequency of graft rejection or survival. Additional data is required before any strong conclusions can be made.

SCS patients

	Absolute Change	Change in SDS
Height change	+3.8cm	+0.40
Weight Change	+4.4Kg	+0.57

LCS patients

	Absolute Change	Change in SDS
Height	+5.85cm	+0.23
Weight	+6.8Kg	+0.54

Title: Vitamin D status in paediatric kidney transplant recipients

S Saleh, NJ Webb, ND Plant, M Shenoy

Objective: To determine the Vitamin D (25 hydroxycholecalciferol) levels and assess response to treatment with a single large dose of Vitamin D2 (Ergocalciferol) in children following kidney transplantation.

Methods: Vitamin D and parathyroid hormone (PTH) levels were measured in all 58 (17 female) children attending the Transplant Clinic at Royal Manchester Children's Hospital between December 2010 and February 2011. Children who were deficient were treated with a single oral dose of Ergocalciferol (<5yr 60,000 units, 5-10yr 100,000 units and >10yrs 160,000 units). Vitamin D and PTH levels were again measured 10-14 weeks after treatment.

Results: The mean age of the group was 14 years (39 Caucasian, 18 Asian and 1 of Afro-caribbean origin. 49 (84.4%) had less than the recommended Vitamin D level of >30ng/ml. Severe VDD (<10ng/ml) was present in 44.8% and insufficiency (10-20ng/ml) was present in 51%. Majority of these children showed evidence of hyperparathyroidism. Following treatment with a single dose of Vitamin D2 (29 children), although PTH levels normalized in the majority of children, only 41.3% had normal levels of Vitamin D.

Conclusion: VDD is common in paediatric renal transplant recipients. A single dose of Vitamin D2 may not be sufficient to correct VDD in the majority of patients.