General paediatrics

**G160** PREVALENCE AND DISTRIBUTION OF PETECHIAE IN WELL INFANTS

A.J. Downes, D.S. Crossland, A.F. Mellon. Department of Paediatrics, Sunderland Royal Hospital, Sunderland, UK

**Introduction:** The presence of a non-blanching rash is an important sign in meningococcal disease. In order to determine the specificity of petechiae in unwell children as a marker for disease the prevalence and distribution in the normal population needs to be established. The published literature says little about the presence of such petechiae.

**Aims:** To determine the prevalence and number of petechiae in well infants.

**Methods:** One hundred and sixteen apparently healthy infants attending child health clinics for surveillance or immunisation purposes were fully examined between October 2000 and February 2001. Three families refused consent. The number and site of any petechiae were recorded.

**Results:** Thirty two babies (27.6% 95% confidence interval [CI] 19.7% to 36.7%) had one or more petechiae but in only 4 cases had these been previously noted by the carer. Ten infants (8.6%) had two or more petechiae (95% CI 4.2% to 15.3%) and three (2.6%) had more than two (95% CI 0.5% to 7.4%). Nine infants had petechiae above the nipple line, 18 below the nipple line and 5 in both areas. None of these babies subsequently developed sepsis.

**Conclusions:** One or two petechial spots are commonly found in healthy infants, their presence alone should not be considered pathological without other clinical signs. In well infants petechiae are more commonly found below the nipple line.

**G161** WHAT ARE THE CAUSES OF A NON-BLANCHING RASH IN CHILDREN PRESENTING TO A PAEDIATRIC EMERGENCY DEPARTMENT: A PROSPECTIVE STUDY

J.C. Smith, L.C. Wells, V.C. Weston, J. Collier, N. Rutter. Queens Medical Centre, Nottingham, UK

**Objectives:** to define the causes of a non-blanching rash in children and to examine the influence of age and season.

**Design:** a prospective cohort study.

**Subjects:** 233 infants and children up to 15 years of age presenting with a non-blanching rash over a twelve month period.

**Setting:** the children’s accident and emergency department of a large teaching hospital.

**Method:** clinical features and laboratory investigations were recorded at presentation and analysed.

**Results:** 87 (37%) had an identifiable cause for their rash. 58 (25%) were directly related to a mechanical cause, 28 (12%) had meningococcal disease, 16 (7%) had a streptococcal tonsillitis, 29 (12%) had an identified viral cause and 15 (6%) had a haematological disorder. Viral infection and meningococcal disease were more common in pre-school children, streptococcal throat infections more common in school children.

Mechanical causes were seen mainly in winter and spring (73%) as was streptococcal throat infection (81%). 50% of meningococcal disease presented in the winter. Respiratory viral infections peaked in the winter, gastroenteral viral infections in the summer.

Those with mechanical causes for their rash, a previously little described group, were mainly pre-school children presenting with either coughing, vomiting or trauma (93%). 46% had a fever (>37.5°C, 95% had petechiae alone lesions<2mm) with no purpura, 64% had a rash confined to the SVC distribution and the remaining 36% had a trauma-related rash. Although most were well, 88% were admitted (97% for 48 hours or less).

**Conclusions:** There are several identifiable causes for a non-blanching rash in child. Some have a viral or minor bacterial infection and a minority have meningococcal disease. Mechanical petechiae from coughing, vomiting, or trauma are common.

**G162** DOES PARENTAL REPORTING OF SMELLY URINE INDICATE URINARY TRACT INFECTION IN YOUNG CHILDREN?

S. Struthers, J. Scalon, K. Parker, R. Hallett. Department of Paediatrics, St. Mary’s Hospital, Milton Road, Portsmouth, UK

**Background:** Parents often report that young children have “smelly urine” or a particular odour. There is little evidence that these observations are relevant to the diagnosis of urinary tract infection (UTI).

**Aim:** To determine whether parental reporting of smelly urine is of any relevance to the diagnosis of UTI in children less than 6 years of age.

**Design of Study:** Analytical, cross-sectional study.

**Setting:** the paediatric admissions unit of a large district hospital.

**Methods:** Parents whose children were having urine collected as part of their assessment were given a simple questionnaire to complete regarding the current smell of their child’s urine. Parents were asked whether their child’s urine smelled different from usual or had a particular smell. Microscopy and culture results of the child’s urine were compared to their parent’s questionnaire answers to see if there was a correlation between parental reporting of different or particular urinary smell and a diagnosis of UTI. Predictive values, specificities, and specificities were calculated for each questionnaire answer.

**Results:** One hundred and ten questionnaires and urine samples were obtained. Fifty two percent of parents thought that their child’s urine smelled different from usual or had a particular smell. Only 6.4% of children were diagnosed as having a UTI. There was no statistically significant association between parental reporting of abnormal urine smell and diagnosis of UTI (p=0.709).

**Conclusion:** In determining whether a young child has a UTI, asking parents about urine smell is unlikely to be of benefit.

**G163** DETECTION OF SERIOUS ILLNESS IN ACUTE PAEDIATRIC ADMISSIONS AFTER A LIMITED PERIOD OF OBSERVATION

P.J. Powell, R. Arya. Department of Child Health, The Royal Bolton Hospital, UK

**Aims:** To evaluate the minimum period of observation necessary to detect serious illness (SI) in acute paediatric admissions in a district general hospital setting.

**Methods:** Analysis of observations made on 250 consecutive acute admissions to a DGH. Serious illness was defined as the need for intravenous fluid or antibiotic treatment, administration of oxygen, acute nasogastric-tube feeding or the presence of an unstable neurological state. Nursing assessments and records of temperature, pulse, respiratory rate and the presence or absence of a non-blanching rash were made at the point of admission and at least at 4 hourly intervals from then. The time at which SI became evident was recorded. The positive and negative predictive value of single measurements or combinations of physiological abnormalities were calculated.

**Results:** 64 (26%) of the 250 admissions had SI during their admission. This was apparent by 4, 8, and 12 hours for 86%, 95%, and 100% of the cohort. Increasing numbers of physiological abnormality were associated with the increasing likelihood of being SI, with 100% prediction of SI for more than 3 such abnormals, 86% for 2 abnormals, and 100% for 1 abnormal. However although specificity for SI was high (82% for pulse rate; 93% for temperature) sensitivity and positive predictive values were low for all such recordings. Absence of physiological abnormality at 4 hours is strongly predictive of a lack of SI.

**Conclusions:** Observation of undifferentiated acute paediatric admissions for a period of 4 hours will enable sufficient data to be acquired to predict whether or not a child will be seriously ill in over 90% of cases. This may enable safe discharge after a short period of observation for up to 74% of acute paediatric referrals to DGHs.

**G164** DEATHS DUE TO INVASIVE PNEUMOCOCCAL DISEASE (IPD) IN ENGLAND AND WALES–A MODEL FOR ESTIMATING EXCESS MORTALITY

E.D.G. McIntosh1, R. Booy2 1Wyeth Vaccines, Taplow SL6 0PH, UK; 2Barts and the London School of Medicine and Dentistry, Queen Mary, University of London, Whitechapel E1 1BB, UK

**Aim:** In the USA, IPD mortality may be underestimated by between 1.5 and 45%. Our aim was to estimate the proportion of “unspecified” mortality in infants and young children aged 1 month to 4 years as reported by the Office for National Statistics (ONS) using ICDP code for England and Wales that could reasonably be attributed to IPD.

**Methods:** Bacterial culture has been the “gold-standard” for the detection of Streptococcus pneumoniae in IPD. More sensitive tests such as the polymerase chain reaction (PCR) have been assessed in paediatric populations. We theoretically applied the results of such tests to ONS “unspecified” mortality due to meningitis, septicaemia,
G165 INVASIVE GROUP A STREPTOCOCCAL INFECTION—SERIOUS YET POORLY RECOGNISED
P. Salt, C. Wynne, S. Nadel, J. Britto. Paediatric Intensive Care Unit, St Mary's Hospital, Paddington, London

Background: The incidence of invasive GAS infection although uncommon, appears to be increasing and carries significant risk of morbidity and mortality. However it is not well described in standard paediatric literature.

Aims: To define presenting characteristics and clinical course of invasive GAS infection on a Paediatric Intensive Care Unit (PICU).

Method: Retrospective, descriptive review of all children admitted to PICU from January 1994 to October 2001 with invasive GAS infection. Data was collected regarding age and sex, presenting symptoms, infection site and duration, duration of stay of gas infusion, length of stay of assisted ventilation, presence of shock and multiorgan failure and outcome.

Results: Deaths due to paediatric ip in the UK may be underestimated by up to 67%. This methodology provides a model for estimating the excess mortality due to under-reported diseases.


G166 DOES REMOTENESS FROM ACUTE SERVICES INFLUENCE ACUTE MEDICAL ADMISSIONS IN INFANTS AND YOUNG CHILDREN?
W. Zaw, J. McDonald, P. Helms. Department of Paediatrics, Raigmore Hospital, Inverness, and Department of Child Health, University of Aberdeen, Scotland, UK

Aims: Accessibility to acute paediatric services is a sensitive political issue and may modify referral and discharge practice. We therefore reviewed the influence of accessibility on referral to and length of hospital stay for acute illness in infants and young children less than 2 years of age.

Methods: All acute admissions between 1 January 1995 and 31 December 1999 to a single acute paediatric unit in Highland Region (Raigmore Hospital, Inverness) were reviewed. Accessibility was defined as mean travel time (minutes) by road from the largest population centre in each of eight clearly defined sub-regions. Average annual admission rates were calculated per 1,000 children less than 2 years of age in each sub-region.

Results: During the study period, there were 3,046 acute medical admissions in children aged 0—2 years. There was a significant negative association between travel time and admission rate (Spearman rank correlation coefficient = 0.83; p<0.01) but there was no significant association between travel time and length of hospital stay (Spearman rank correlation coefficient = 0.04; p=0.38). In multiple regression with adjustment for regional socio-economic deprivation (proportion of adults receiving income support) the regression coefficient for travel time remained statistically significant (β=—0.321; p<0.028).

Conclusions: In a predominantly rural region and up to a 2-hour travelling time, remoteness from acute paediatric services is associated with lower admission rates of young children and infants with acute medical problems.

G167 HOME CARE FOR SICK CHILDREN—AN UNDERDEVELOPED SERVICE?
C.E. Cramp. Royal Shrewsbury Hospital NHS Trust, UK

Aim: To establish the extent of current facilities in the UK for the home care of acutely sick children—“Hospital at Home”—which could replace all or part of episodes of inpatient care.

Method: A questionnaire was sent out from RCPCH in December 2000 to all paediatric clinical directors listed on the College database.

Results: Responses were received from 153 (out of 301) trusts—66 acute units, 44 combined acute/community units and 26 community units. 95 units run home nursing services from their trust, allowing identification of 3 groups:

1. Biased towards acute care: 19 (23%) units - mean of 991 children managed at home each year—acute cases 68—average length of episode of care 1—10 days—high turnover (203 children managed/nurse/year)—70% services available in evenings and weekends, some available at all times.
2. Divided between chronic and acute on chronic illness: 46 units—mean of 572 children managed each year—average episode weeks/months—acute cases 32—moderate turnover (130 children managed/nurse/year).
3. Entirely chronic illness: 10 (12%) units—continuing care/episodes for a mean of 144 children/year—low turnover (48 children managed/nurse/year)—mainly small services available in the daytime only.

Impact on inpatient services varied according to the type of team. For mainly acute teams, 41% referrals to home care were replacements for inpatient care, while 37% replaced outpatient appointments or ward attendances. For acute/chronic teams, these percentages were 31% and 62% and for chronic teams, 30% and 34% respectively. Impact, especially on inpatient care, was higher for services available at nights and weekends.

Conclusions: Home nursing care can replace inpatient care for some children. Acute episodes are less costly in nursing time than chronic care and are more likely to replace inpatient provision. Many trusts currently have no access to home nursing for acute illness. The use of home nursing services for acute episodes of illness has the potential for considerable further development with reduction in the use of inpatient facilities.

G168 TWO CENTRE COMPARISON OF THE MANAGEMENT OF BRONCHIOLITIS ASSOCIATED APNOEA
M.P. Samuels, A. Al-Balkhi, K. Marinaki, D.A. Thomas, H. Klonin, D.P. Southall. Queen’s Medical Centre, Nottingham and North Staffordshire Hospital, Stoke-on-Trent, UK

Aims: Apnoea in bronchiolitis commonly results in intubation and positive pressure ventilation (PPV). Nasal CPAP and negative extrathoracic pressure (NPV) have also been used to avoid intubation. We examined the frequency of use of PPV in patients with bronchiolitis associated apnoea cared for in two PICUs, one of which offered NPV.

Methods: Case notes review of all patients referred from 10/93 to 2/97 with a clinical diagnosis of bronchiolitis associated apnoea.

Results: There were 31 (77% RSV+ve) admitted to the NPV centre and 21 (86% RSV+ve) to the centre offering only PPV. There were no significant differences between the two units for the following patient details: age, sex, weight, birth weight or gestation, history of apnoea
of prematurity, days ill before presentation, oxygen requirement at ICU referral, or proportion transferred from other hospitals. There were no significant differences in patient management and outcome:

<table>
<thead>
<tr>
<th>Abstract G168</th>
<th>Management / outcome</th>
<th>PPV-centre</th>
<th>NPV-centre</th>
</tr>
</thead>
<tbody>
<tr>
<td>No of patients admitted</td>
<td>21</td>
<td>31</td>
<td></td>
</tr>
<tr>
<td>Number receiving NPV</td>
<td>0</td>
<td>23</td>
<td></td>
</tr>
<tr>
<td>Number intubated (%)</td>
<td>(26) 18</td>
<td>(26) 8 *</td>
<td></td>
</tr>
<tr>
<td>Median PICU stay (d)</td>
<td>7</td>
<td>2 *</td>
<td></td>
</tr>
<tr>
<td>*p=0.0001</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

NPV was used safely and effectively during retrieval of 13 patients from other hospitals.

Conclusions: In these two groups, the use of NPV was associated with less need for PPV and PICU stay. Negative pressure ventilation should be considered in the treatment of bronchiolitis associated apnoea.

G169 USE OF INTRAVENOUS SALBUTAMOL (IVS) IN CHILDREN WITH ACUTE SEVERE ASTHMA: HOW BAD IS THE CONFUSION?

A. Rao, T. Vigneswaran, P. Ramnarayan, G. Roberts, J. Britto. Paediatric Intensive Care Unit, St Mary’s Hospital, Paddington, London

Background: In response to accumulating evidence, several national guidelines have recently suggested the use of intravenous salbutamol (IVS) in children with acute severe asthma. However, British Thoracic Society guidelines (1997) do not indicate any role for its use. Have these differences caused significant heterogeneity in the management of children with asthma?

Aims: 1. To describe the use of IVS in children with acute severe asthma prior to referral for intensive care 2. To identify the role of IVS in asthma protocols across hospitals in the South-East of England

Methods: Data were collected on all children with acute severe asthma referred to a paediatric intensive care unit (PICU) from Sept 1999–Oct 2001. IVS use at the referring hospital, dosing practice, concurrent use with aminophylline and outcome of the referral were recorded. Paediatricians were contacted by telephone in each hospital that referred patients to our PICU in order to establish the use of IVS in their current asthma guidelines and a facsimile copy was obtained.

Results: Out of 51 children on whom data was available, 25 received IVS prior to being referred for intensive care (group 1), a further 23 only after discussion with PICU (group 2). In group 1, most (23/25) children had already received a trial of aminophylline. Only 11 had a bolus dose of IVS (mean 9.6 mcg/kg) and 20 were managed on infusions (mean starting infusion rate 2.7 mcg/kg/min). 19/25 (76%) children from group 1 and 13/23 (57%) from group 2 were subsequently transferred to PICU. 30 out of 61 hospitals contacted used IVS in acute asthma. Most hospitals used it after a trial of aminophylline; only 11 used it as the intravenous bronchodilator of choice. There was widespread variation in dosage.

Conclusions: Lack of consensus between national guidelines has resulted in considerable variation in the use of intravenous salbutamol. The role of this drug in acute asthma needs urgent clarification in order to ensure uniformity in management.

G170 CAN ADENOTONSILLECTOMY BE AVOIDED WITH MEDICAL TREATMENT?

A.J. Wheeler, V. van Someren. Royal Free Hospital, London

Aims: To determine if nasal steroids are therapeutic in children on the waiting list for adenotonsillectomy with recurrent tonsillitis (RT) and/or obstructive upper airway symptoms (OS).

Methods: A double blind, placebo controlled trial of four months treatment with nasal fluticasone (200mg/day) prior to adenotonsillectomy in children aged from 4 to 12 years (n=84). Primary outcome measures were defined as a 40% improvement in symptom scores, improvement in sleep study results and cancellation of surgery due to improvement in symptoms. A follow-on immunohistochemistry study was undertaken to identify if there was a steroid effect at the cellular level.

In this study lymphocyte activation phenotype of tonsil and adenoidal cells were determined using flow cytometry.

Results: Nasal steroids were not found to benefit children on the waiting list for adenotonsillectomy in terms of the primary outcome measures listed. However subgroup analysis of the steroid treated group revealed the following profile: RT alone had a 50% increase in symptoms (p<0.03); OS alone had a 40% improvement in symptoms (p<0.05); RT and OS together had a mixed picture with no change in symptoms. The immunology results showed that there was a difference in adenoidal T cell phenotype with steroid treatment: an increase in naive T cells (CD45RA+, p=0.04) and a reduction in activated T cells (CD45RO+, p=0.001 and HLA-DR, p=0.02). These are effects that would be predicted with steroid treatment. No such differences were seen in tonsil lymphocyte populations.

Conclusions: As the subjects in the subgroup with OS alone had the same prevalence of otitis as compared with the control group, the improvement in OS is likely to be due to a reduction in adenoidal hypertrophy. The immunology results add weight to this conclusion as they suggest that topical steroids do penetrate the adenoids, reaching a tissue concentration that has an expected pharmacological effect. Thus for children with OS alone nasal steroids may relieve symptoms and for some surgery could be avoided.

G171 DOSE-RESPONSE RELATIONSHIP OF OCCLUSION THERAPY FOR AMBLYOPIA: RESULTS FROM THE MONITORED OCCLUSION TREATMENT FOR AMBLYOPIA STUDY (MOTAS)

C.E. Stewart, A.R. Fielder, M.J. Moseley, D.A. Stephens for the MOTAS Cooperative group. Department of Ophthalmology, Imperial College of Science, Technology and Medicine, Western Eye Hospital, 171 Marylebone Road, London, NW1 5QH

Introduction: Amblyopia is a common visual disorder affecting 3–5% of children. Occlusion therapy is the mainstay treatment, yet neither the effectiveness nor the time course of this treatment is known. Though prescribed occlusion dose may exceed 3,000 hours.

Aims: To determine the kinetics of the dose-response relationship for occlusion therapy.

Methods: Data were obtained from 57 subjects (mean age 5.1 ± 1.4 years) with amblyopia associated with strabismus (n=22), anisometropia (n=13), and with both anisometropia and strabismus (n=20). Forty-eight subjects required refractive correction and wore spectacles for 18 weeks before starting occlusion. All subjects were prescribed 6 hours occlusion/day. Outcome variables (logMAR visual acuity, log contrast sensitivity and stereoaucuity) were assessed at two weekly intervals until gains in visual acuity ceased to be statistically verifiable. Occlusion was objectively recorded using an occlusion dose monitor (Fielder AR, et al. Lancet, 1994;343:547).

Results: Mean visual acuities at onset and end of occlusion were 0.53 (6/19) ± 0.36 and 0.26 (6/11) ± 0.16 logMAR respectively. Approximately 85% of visual acuity improvement occurred in the first 6 weeks of occlusion. Children ≤ 5 years showed a greater improvement (change in VA; 0.39 log units (4 chart lines) vs. 0.12 log units (1 chart line)) than children > 5 years (P<0.01). The relationship between visual acuity gain and total occlusion dose is described by a monotonic function, which for all categories of amblyopia appears to be linear up to 160 hours of total recorded dose.

Conclusions: Occlusion therapy is most effective in the first few weeks of treatment with outcome positively correlated with dose and age.

G172 HAVE FASTING GUIDELINES REDUCED FASTING TIMES IN PAEDIATRIC SURGICAL PATIENTS?

S. Walsh, C.A. Ryan, G. Shorten. Department of Paediatrics & Child Health and Department of Anaesthesia, Cork University Hospital, Cork

Background: Traditionally despite their vulnerable physiological children have been made to fast similar to adults from midnight prior to elective surgical procedures. Following studies which showed that gastric emptying time in children for clear fluids was 20 minutes, and that clear fluids may in fact accelerate gastric emptying.

Shorter fasting times for children were recommended in 1993. These guidelines specify fasting times of 2 hours for clear liquids and 6 hours for solids for children >1 year. The object of this study was to compare fasting times in three different paediatric surgical centres (two in Cork and one in France).

Methods: Fasting times on surgical patients in paediatric wards were measured in 3 centres, one in France and two in Cork. Fasting
Introduction: Paediatric inpatient prescribing has been computerised in our department for the last 12 years. In 2001 the pathway for prescribing 14 common paediatric medicines was changed from traditional clinician calculation (‘main pharmacy’ pathway) to directed weight and age-banded prescribing (‘paediatric’ pathway). Individual prescribers can still opt to use either the ‘main’ or ‘paediatric’ pharmacy pathways for each medicine.

Aim: To determine whether the ‘paediatric’ prescribing pathway has reduced prescribing errors.

Method: All paediatric inpatient prescriptions were prospectively collected over a 5 day period and divided into those prescribed via the ‘paediatric’ pathway, and the ‘main pharmacy’ pathway, as well as identifying the prescribers as paediatricians or non-paediatricians (surgeons). Prescriptions were analysed, and results presented as error rates.

Results: 160 children were studied (FP 53; CP 54; CO 53). Mean fasting time for liquids in some instances up to 6 times longer than and mean fasting time for solids were more than double the recommended guideline levels (see figure). The hospitals with previously instituted guidelines (CP and FP) had significantly shorter fasting time compared to the Hospital CO without guidelines (11.2hrs V 12.9hrs for liquids, 13.2 hrs V 14.7 hrs for solids). Prolonged fasting from liquids (greater than 15 hours) was more common in both Cork Units (CO 27%, CP 29%) compared to the French Unit (9%) (see figure). The hospitals with previously instituted guidelines (CP and FP) had significantly shorter fasting time compared to the Hospital CO without guidelines (11.2hrs V 12.9hrs for liquids, 13.2 hrs V 14.7 hrs for solids).

Conclusion: The introduction of fasting guidelines has been associated with mean reduction in fasting times. Worryingly however, over a quarter of all paediatric patients undergoing elective surgery in the Cork Hospitals still fasted for more than 15 hours.

G173 COMPUTERISED WEIGHT AND AGE-BANDED PRESCRIBING REDUCES PAEDIATRIC INPATIENT ERRORS IN A DISTRICT GENERAL HOSPITAL

J.L. Robertson, N.A. Caldwell, W. Roberts, B. Power, A.P. Hughes. Paediatric Department, Wirral Hospital NHS Trust, Upton, Wirral, CH49 5PE, UK

Aims: To determine the incidence and causes of SH in infancy. To identify any problems in management and hence improve practice guidelines.

Patients and Methods: Case note audit of infants admitted with SH, to St George’s Hospital, from Jan 1995–Dec 2000. 8 cases were identified, 7 presented with seizures, 1 in cardiac failure. Median age was 15.5 days (range 7–126).

Results: VDD was the cause of SH in all cases. All mothers came from at risk groups, none had received pre or post-natal advice on supplementation. All tested mothers had very low 25-OHVD levels. 1 mother refused blood tests. 6/7 had raised PTH levels despite normal
bone profiles in all. 25-OHVD was low in all but 1 infant, in whom it was sent post 1alfacalcidol treatment. 4 infants, who presented early (aged 7–10 days) had normal/high PO4 and low PTH levels. This suggested an initial diagnosis of primary hypoparathyroidism, resulting in further investigations for associated disorders. The hypoparathyroidism was transient resolving with VD treatment. The 4 infants who presented later (aged 21–126 days) had high PO4, and raised PTH levels, suggesting PTH resistance. This was transient resolving with VD treatment. A low Mg²⁺ was seen in 7 infants. Intravenous Ca²⁺ and Mg²⁺ boluses were felt to be warranted in 6 and 5 infants respectively. Intravenous Ca²⁺ infusion was continued in 3 infants, resulting in 1 significant extravasation injury. Diagnostic uncertainty meant that VD treatment was delayed until day 3 in 4 infants and day 15 in one.

Conclusions: VDD leading to SH in infancy still occurs, as antenatal supplements are not universally given in all at risk groups. Hypomagnesaemia, transient hypoparathyroidism and PTH resistance are all seen in VDD. These different biochemical profiles should be incorporated into guidelines for management of SH in infancy. 1alfacalcidol treatment should be started in infants with SH, at presentation, pending confirmation of diagnosis. Ca²⁺ infusions should be stopped as soon as practicable, limiting their use to persistently symptomatic infants and/or those not tolerating oral treatment.

NAIL PATELLA SYNDROME: MUCH MORE THAN A SKELETAL DYSPLASIA

E. Sweeney, A.E. Fryer, R.C. Mountford, A.J. Green, I. McIntosh. Mersey-side and Cheshire Clinical Genetics Service, Royal Liverpool Children's Hospital, Alder Hey, Liverpool L12 2AP; Merseyside and Cheshire Regional Molecular Genetics Laboratory, Crown Street, Liverpool, L8 7SS

Aims: To determine the spectrum, incidence and severity of features associated with Nail Patella Syndrome (NPS). To identify mutations in the NPS gene, LMX1B.

Methods: Interview, examination and urinalysis of 123 British NPS patients and review of case notes and x rays. Mutation detection by SSCP and direct sequencing.

Results: 98% of patients had fingernail changes. 75% of patellae were hypoplastic and 9% absent. There was loss of extension in 70% of elbows and 12% of patients had pterygia. 19% had congenital talipes. 65% had iliac horns on x ray. Renal involvement was present in 25% of patients (33% in > age 40) with an average age at detection of 22 years (1–51). Only 2% had developed renal failure. 7% had ocular hypertension and 10% had glaucoma (12% and 17% in > age 40) with an average age at detection of 48 years (23–78). Involvement of the GI tract was suggested in 31% of patients by irritable bowel syndrome or constipation. 25% described peripheral neurological symptoms and 7% had epilepsy. Mutations of LMX1B were detected in 27/33 families including 16 novel mutations. There were 11 missense, 8 potential splice site and 4 frameshift mutations, consistent with the hypothesis that NPS is the result of haploinsufficiency for LMX1B.

Conclusions: Along with the skeletal, renal and ocular features seen in NPS, we suggest that gastrointestinal, neurological and vascular symptoms are also part of the NPS phenotype. The broad range of symptoms than classically reported for NPS is in agreement with the pattern of LMX1B expression.

DELAY IN DETECTION OF CLEFT PALATE, AN AUDIT OF NEWBORN CLINICAL EXAMINATION

A. Habel1, N. Elhadi2, B. Sommerlad1. 1 Great Ormond Street Hospital for Children, London; 2 West Middlesex University Hospital, Isleworth

Aims: Examination of the newborn is done to detect manifest abnormalities. It was evident that a proportion of patients with open cleft palate (CP) presenting to a single referral centre for cleft lip and palate surgery were not identified at that first newborn examination. An audit of the newborn was performed to determine the spectrum, incidence and severity of features associated with NPS. To identify mutations in the NPS gene, LMX1B.

Methods: All referrals found to have CP from 1990 to 2000 were subjected to case note review and a questionnaire was sent to the parents asking for their recall of symptoms such as feeding and breathing difficulties.

Results: 317 individuals were identified. 65% were CP. 35% were submucous cleft palate and were not considered further for this report. 37% of CP were isolated (ICP), 63% had other malformations or syndromes (SCP).

Among ICP 69% were detected within the first 24 hours, 13% the rest of first week, 8% before 2 months and 10% after 2 months (up to 5 years).

In SCP the presence of more than one anomaly improved detection, but not significantly; first 24 hours was 78%, rest of first week 15%, before 2 months 3%, after 2 months 4%.

The size of the cleft, soft only or soft and extending into the hard palate, was not related to delayed detection.

109 CP questionnaires were returned. Feeding difficulties and/or regurgitation of feed was present in all those with delayed detection.

Conclusion: Failure to identify a cleft in 31% with ICP and 22% with SCP in the first 24 hours was found. Associated symptoms were reportedly common before detection.

Improved detection would result from the routine use of direct visualisation of the palate with a spatula and torch.

ABDOMINAL PAIN, APLEY REVISITED

W. ElMatary, C. Spray, B.K. Sandhu. Institute of Child Health, Bristol, UK

Aims: The aims of the study were to: 1) Revisit Apley using modern methodology 2) Prospectively assess children with RAP and identify children not associated with organic and non-organic pain.

Methods: 317 individuals were identified. 65% were CP. 35% were ICP. 109 CP questionnaires were returned. Feeding difficulties and/or regurgitation of feed were present in all those with delayed detection.

Conclusion: Failure to identify a cleft in 31% with ICP and 22% with SCP in the first 24 hours was found. Associated symptoms were reportedly common before detection.

Improved detection would result from the routine use of direct visualisation of the palate with a spatula and torch.
scores ranged from -1.63 to 14.5. Data were analysed using the Kruskal-Wallis Test; a non-parametric analysis of variance.

No significant differences were seen between the three groups in age and sex distribution or distribution of families in relation to Townsend scores. Our data suggest there is a reluctance among families for their children to participate in a clinical trial involving some investigations and recruitment was not influenced by socio-economic status.