Neurology

G177 THE CONTINUING ROLE OF TENSILON (EDROPHONIUM) TESTS IN THE DIAGNOSIS OF CHILDHOOD MYASTHENIA

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Aim: Myasthenia is a rare paediatric disease. Traditionally, the Tensilon test, neurophysiological (NP) studies and serology have been used in its diagnosis. This first study in children was to clarify the role and safety of the Tensilon test in the initial diagnoses of myasthenia when compared with, and in association with NP studies such as stimulated single fibre EMG (sSFEMG) and repetitive nerve stimulation (RNS).
Methods: Retrospective non-comparative case note review of 44 patients who have had Tensilon tests requested in the last 10 years for the diagnosis of myasthenia in a tertiary paediatric neurology centre.
Results: 37 Tensilon tests were carried out as per local hospital protocol. Seven of 44 patients did not have the Tensilon test carried out because of a change in clinical status. 34 of 37 children had ocular symptoms at the time of the Tensilon test. 33 of 37 children had Tensilon test and NP studies (sSFEMG or RNS or both). In 26 of 33 the results of Tensilon test and the NP studies were concordant. 13 of 26 patients had both tests suggestive of a defect in the neuromuscular junction. All these patients had a final diagnosis of myasthenia (autoimmune myasthenia (n = 7), congenital myasthenia (n = 5)). The remaining 13 of 26 children had both tests negative and did not have a final diagnosis of myasthenia. Seven of 33 children had discordant results. Five of seven discordant test group had a positive Tensilon test and normal NP studies. The final diagnosis was autoimmune myasthenia (n = 3), seronegative myasthenia (n = 1) and mitochondrial disease (n = 1). Two of seven patients had a negative Tensilon test and positive NP studies. The final diagnosis was congenital myasthenia (n = 1) and seronegative myasthenia (n = 1). Five of 37 children had minor and transient side-effects after Tensilon was injected. None of the children required atropine or had severe adverse effects.
Conclusions: The Tensilon test continues to be a safe bedside test (when performed as per standardised protocol) with minimal complications. During initial presentation especially in myasthenia with ocular symptoms, NP studies could be normal and at that stage, a Tensilon test would be advisable. A concordant test with both Tensilon test and NP studies can confidently diagnose or rule out myasthenia.

G178 SINGLE PHOTON EMISSION COMPUTED TOMOGRAPHY SCAN IN PRE-SURGICAL EVALUATION OF CHILDREN WITH REFRACTORY EPILEPSY

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Introduction: Single photon emission computed tomography (SPECT) is a nuclear medicine imaging technique that uses a radioactive pharmaceutical, injected during a seizure to identify an epileptogenic focus seen as a hyperintensity on the ictal scan. The aim of the study is to define the role of SPECT study in the presurgical evaluation of children with refractory epilepsy.
Methods: This is a retrospective study from years 2002 to 2007. All the patients who underwent an ictal SPECT scan for presurgical evaluation were identified and then the results of the MRI scan, EEG and SPECT scan from this group of children were analysed. The role of the SPECT scan in this process of decision making was analysed.
Results: A total of 172 children were identified. SPECT scan localised a possible epileptogenic focus in a total of 85 (49.4%) children, showed hemispheric lateralisation in 30 children and was non-localising/lateralising in 57 children. 89 (51.74%) children were considered suitable for epilepsy surgery of which 54 (60%) children had localised SPECT scan. 75 of 89 children had unihemisperic structural abnormality with 48 patients with a localised SPECT scan. 27 children (including one with hypothalamic hamartoma) with bilateral imaging abnormality were evaluated during this period and 12 (44.5%) were offered surgery. All children offered surgery had a concordant EEG, while eight had a localised or lateralised SPECT scan in the group.
Conclusions: Ictal SPECT has a role, in concordance with EEG in pre-surgical evaluation for children with bilateral imaging abnormality and for repeat surgery if required.

G179 PRESENTATION AND DIAGNOSIS OF AICARDI–GOUTIÈRES SYNDROME IN INFANTS: A CASE SERIES

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Aicardi–Goutières syndrome (AGS) is a rare inherited encephalopathy caused by mutations in the AGS1 (*TREX1*) or any one of at least five genes, labelled AGS2–6. We present 11 cases (from eight families) of genetically confirmed AGS presenting to a paediatric neurology unit. Common presenting features included developmental delay or regression (n = 11), microcephaly (n = 11), hypotonia leading to dystonia and spasticity (n = 11), epilepsy (n = 11) and ''chilblain-like'' skin lesions (n = 3). Unusual presenting features included hypocalcaemia (n = 1), thrombocytopenia (n = 1), haemolytic anaemia (n = 1) and opsoclonus myoclonus (n = 1). All had calcification on neuroimaging. One child did not have raised interferon α or white blood cells in their CSF, but did have raised CSF pterins. Of the eight families, four were positive for the *TREX1* mutation, two had a mutation in the AGS2 (RNaseH2) gene and one patient had a mutation in the AGS3 gene. A further patient fit the clinical phenotype of AGS and had raised CSF interferon α, but has no mutations in the AGS1–5 genes. All the *TREX1* mutations presented within 2 months of life with features similar to congenital infection. All were of South Asian origin. The AGS2 and 3 mutations presented between birth and 8 months of life. This paper highlights the importance of genetic testing in the diagnosis of AGS and raises the possibility of ethnic diversity in the inheritance of the *TREX1* gene mutation.

G180 THE MANAGEMENT OF MENTAL HEALTH NEEDS OF CHILDREN WITH EPILEPSY ATTENDING A PAEDIATRIC NEUROSCIENCES SERVICE

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Introduction: Recent guidelines (UK National Institute for Clinical Excellence and Scottish Intercollegiate Guidelines Network (SIGN)) indicate the high levels of mental health needs in children and young people with epilepsy. Mental health needs may be identified and access to appropriate services facilitated by close working between paediatric and mental health professionals.
Aim: To describe services supporting the mental health of children and young people with epilepsy attending a multidisciplinary specialist regional neurosciences service with a range of resources available and an established psychosocial epilepsy research theme. Detailed attention is given to presentations where the interplay of epilepsy and cognitive, behavioural, emotional and/or psychiatric difficulties is dealt with in neuropsychology and neurology/psychiatry liaison services.
Method: The frequency of referrals of children with epilepsy to a range of community- and hospital-based child and adolescent mental health services (CAMHS) is estimated from practitioners' records.

Files of children with epilepsy open to liaison psychiatry on 1 October 2007 were examined to determine patterns of service involvement.
Results: The paediatric neurosciences service covers a population containing about 2000 children with epilepsy. Some of these are seen by paediatric psychology or community CAMHS. Annually over 100 are referred for a neuropsychology service. Children referred to neuropsychology generally present with cognitive, emotional and behavioural difficulties; a small number with more significant mental disorder are seen jointly with the neuropsychiatrist. In the year 2007–08, over 50 neurosciences referrals to liaison psychiatry were made; epilepsy is the most common neurological diagnosis and an epilepsy diagnosis predicts a longer liaison intervention. 22 such cases were open to the liaison team on 1 October 2007, mean age 12 years (M:F ratio 2:1). The mean duration of liaison team input was 18 months. Most received individual and family work, joint neurology/psychiatry clinics, psychological assessments and education psychology advice; half had social work input. SIGN recommendations regarding the use of psychotropic drugs were met.
Clinical implications: Description of the range of CAMHS utilised by patients of a specialist centre suggests a care pathway model for the management of mental health needs of children with epilepsy.

G181 ELECTROGRAPHIC SEIZURE-LIKE ACTIVITY IN PRETERM INFANTS IN THE FIRST WEEK OF LIFE IS ASSOCIATED WITH CEREBRAL INJURY

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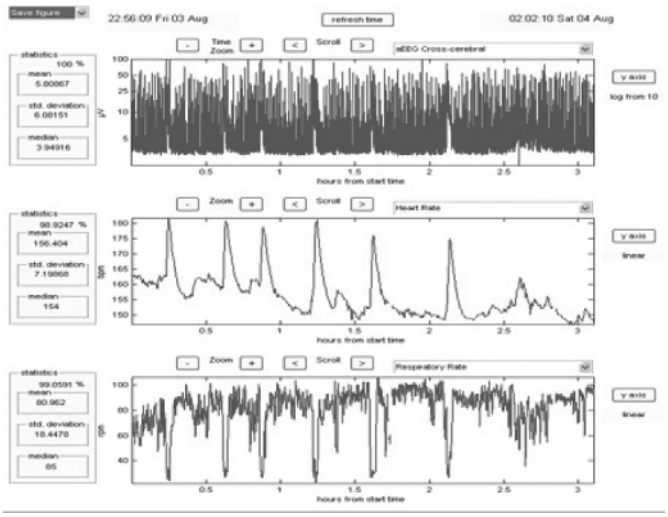
Objective: The aim of this study was to determine the incidence of electrographic seizure-like activity (ESLA) in a prospective cohort of preterm infants, using amplitude-integrated (a)EEG monitoring, and relate it to cerebral injury.
Methods: Infants born <30 weeks received an optimum 74 h period of continuous two-channel EEG with aEEG monitoring in the first week of life. Infants were classified into the abnormal (AB) outcome group if they died in the neonatal period and/or had grade 3–4 intraventricular haemorrhage and/or moderate or severe abnormalities on cerebral MRI. ESLA were identified using the raw EEG trace in combination with the aEEG and were diagnosed when rhythmic spike and/or wave activity was present for at least 10 s on the raw EEG trace.
Results: 51 infants underwent aEEG monitoring in the first week of life. 11 of 51 (22%) infants displayed ESLA. Infants with ESLA were more premature, had lower birth weights and a greater proportion had AB outcomes compared with those without (9 of 11 (82%) vs 8 of 40 (20%); $\chi^2 = 14.8$, Fisher's exact test, $p<0.001$). Three infants with ESLA displayed a concurrent rise in heart rate and one also showed a fall in respiratory rate (fig). Five of the 11 infants had grade 3/4 intraventricular haemorrhage with ESLA preceding ultrasound findings.
Conclusions: ESLA was more likely to occur in the sicker and more premature preterm infants who had abnormal outcomes. ESLA as detected on continuous digital aEEG monitoring was prognostic for poor outcomes in preterm infants. Autonomic changes may assist detection of ESLA in preterm infants.



Abstract G181

G182 LEIGH SYNDROME: A FAMILIAR PHENOTYPE BUT A DISAPPEARING DISEASE?

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Aims: To report on children with Leigh syndrome identified via our national study of progressive intellectual and neurological deterioration (PIND).
Methods: We have now carried out surveillance for over 11 years using the British Paediatric Surveillance Unit surveillance system to identify UK children with PIND. Clinical information about notified cases, obtained by telephone questionnaire or hospital visit, is anonymised and classified by the PIND Study Expert Group of paediatric neurologists.
Results: By July 2008 UK paediatricians had notified 2493 children with suspected PIND. There were 112 children with mitochondrial diseases among whom the biggest subgroup consisted of 15 children (nine male and six female) with Leigh syndrome. These 15 children presented between birth and 3 years 2 months of age. The numbers with the following presenting symptoms/signs were: developmental delay (five), failure to thrive (four), seizures (four), hypotonia (four), nystagmus (two), respiratory distress (two), choreoathetoid movements (one), pigmentary retinopathy (one), cardiomyopathy (one), squint (one), coarctation (one) (many had more than one of these). The relevant investigations were: raised lactate in blood and/or CSF 11; brain MR results: spectroscopy abnormal (two), increased signal in basal ganglia (six), abnormal signal in brainstem (three), abnormal white matter (one), not done (two), normal (one); mitochondrial DNA mutations (two). Abnormalities in respiratory chain enzymes were reported in four cases but were not specific enough for these cases to be placed in a different diagnostic group. Among the 112 children with mitochondrial disease there were 10 children with relatives who had been diagnosed with "Leigh's disease"; however, only three of these 10 index cases were in our Leigh syndrome group.
Conclusions: There is a recognised Leigh phenotype associated with characteristic scan findings that leads to the diagnosis, but cases often present with non-specific features. It seems likely that fewer cases will be placed in this group as more specific diagnoses are made—most index children with a family history of "Leigh's disease" were not in our Leigh syndrome group.
Acknowledgements: Many thanks to all the UK paediatricians who report cases to the PIND Study, the PIND Expert Group, the British Paediatric Surveillance Unit and the English Department of Health for funding the study.

G183 MAGNITUDE OF OCCIPITOFRONTAL HEAD CIRCUMFERENCE GROWTH IMPAIRMENT IN GOOD OUTCOME VERY LOW BIRTH WEIGHT INFANTS AT 2 YEARS CORRECTED AGE RELATES TO THE DEGREE OF PREMATURITY BELOW 30 WEEKS

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Introduction: Human brain growth velocity peaks at term with the last trimester of pregnancy a critical period for brain volume

(BV) and cortical surface area (SA) development. Cerebral morphometric studies showed that the SA to BV scaling exponent is related to gestational age (GA) in a dose-dependent manner. BV is related to occipitofrontal head circumference (OFC) from birth to 3.5 years. We sought to determine if the pattern of postnatal OFC growth in preterm infants at 2 years also demonstrates GA dose-dependency in infants with no or mild disability.

Methods: A prospectively collected anonymised geographical data set of very low birth weight infants (birth weight <1500 g) born in 1993–2002 was examined. OFCs at birth and at 2 years were expressed as standard deviation scores (SDS) calculated using the 1990 British growth references. Of 1850 infants assessed, data from 567 infants were analysed. Exclusions included birth gestation ⩽25 weeks or ⩾33 weeks, birth weight or OFC outside normal range, multiple birth, congenital malformation, and severe or moderate disability at 2 years. Statistics were performed using Statistical Discovery Software (JMP 7.0). GA trends were assessed using one-way ANOVA followed by post hoc tests.

Results: At birth, there was a relationship between GA and OFC-SDS (trend in 95% CI for mean OFC-SDS from −1.0 to −0.48 in 26 weeks infants to −0.28 to −0.18 in 32-week infants (F-ratio 5.99, $p<0.0001$)). At 2 years, there was no relationship between GA and OFC-SDS (95% CI for mean OFC-SDS was −0.86 to −0.64 (F-ratio 1.25, $p = 0.28$)). The difference in OFC-SDS between birth and 2 years was related to GA (F-ratio 3.73, $p = 0.0012$). 32-week infants had significantly less difference compared with all other groups. Using 32-week infants as controls, OFC growth impairment showed GA-dependent ordering, with the worst effect at 26 weeks (significant mean difference compared with a control of −1.1 and −0.63 in 26 weeks and 30 weeks respectively). Similar effects were observed when the analysis was limited to 381 infants with normal outcome.

Conclusion: These data suggest that in infants born ⩽30 weeks gestation, brain development is vulnerable even in those with apparently good outcome and that the degree of prematurity before this GA has a dose-dependent effect on the pattern of accrual in BV. The ordered GA dependency also indicates the potential presence of a peripartum factor, trigger or switch in brain growth.

G184 CLINICAL, BIOCHEMICAL AND IMAGING OUTCOME IN SEVEN CHILDREN WITH MOLYBDENUM COFACTOR DEFICIENCY

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Introduction: Molybdenum cofactor deficiency (MOCOD) is a rare progressive autosomal recessive neurodegenerative disorder. The most common presentation is with intractable neonatal seizures. The phenotype is increasingly varied. The clinical features and prognosis is not dependent on the pattern or the residual enzyme activity. The majority do not survive early childhood.

Case series: We reviewed the clinical, biochemical, MRI, EEG features and outcome of seven children, diagnosed with MOCOD over the last 11 years. There were four females. The age at presentation ranged from 48 h (five), 8 months (one) and 24 months (one). Time to diagnosis was 4 days to 4 years, (median 3 months). The main presenting features included intractable neonatal seizures (five) and global developmental delay (two). One infant in the latter group developed seizures and neuroregression in early childhood following an intercurrent illness while the other child had bilateral upward lens dislocation. The clinical features included axial hypotonia with limb spasticity (five), dystonia (three), dysmorphism (three), moderate to severe global developmental delay (seven) visual impairment (seven).

Biochemistry: High urinary sulphite, purines and sulphocysteine (seven) and low plasma urate (six). Genetic confirmation was available in three.

Imaging: Global white matter swelling/cavitations (five), predominant anterior involvement (three), symmetrical basal ganglia signal changes within pallidal restriction/T1 shortening (four), cortical highlighting (four), cerebellar hypoplasia (three), symmetrical dentate signal changes (one), post fossa arachnoid cyst (three), ventricular dilatation (three) and cerebral atrophy (five). Burst suppression was seen in the neonatal period in three who had EEG.

Outcome: All children are alive, oldest being 11 years. Moderate to severe learning difficulties (seven), motor disorder (six), visual impairment (seven) epilepsy (two) and enteral feeding (five).

Conclusions: The phenotype in MOCOD is broad. In our series, the two with global delay had additional features that prompted a search for aetiology. Early consideration of diagnosis is essential to inform management (including antenatal diagnosis) and prognosis. Further research is needed to delineate the pathophysiological process so that therapeutic avenues (substitution, gene) could be explored.

G185 TRANSCRANIAL DOPPLER AND STROKE IN SOUTH AFRICAN CHILDREN WITH HUMAN IMMUNODEFICIENCY VIRUS

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Aim: To document the ranges of transcranial Doppler (TCD) velocity values in children with HIV with and without stroke.

Background: The annual incidence of stroke in children with HIV is 1.3% and is thought to be related to systemic immunosuppression and secondary infection. However, HIV infection has been implicated as a primary vascular pathogen with a role in structural and functional vascular changes, including vasomotor reactivity and carotid intimal thickness. There is a reduction in middle cerebral artery time averaged mean maximum velocity (TAMMV) in adults with HIV, thought to reflect alterations of cerebral resistance in arterioles related to HIV or asymptomatic opportunistic infection.[1] There is some evidence for interaction between cerebral lateralisation and immune function[2] but this has not been explored in HIV. TCD enables non-invasive detection of vasculopathy and is an ideal screening tool. TAMMV is often high in children with stroke but there are few data in children with HIV. Our hypotheses were (1) in children with HIV infection, intracranial TAMMV would be lower than the normal range for children (50–150 cm/s), and (2) TAMMV would be higher in children with HIV and stroke than in those with HIV without stroke.

Method: After obtaining informed consent/assent, we recorded TAMMV, clinical and laboratory data from 42 children (24 male, median age 7.4; range 2.6–15.5 years) attending a South African HIV clinic in a prospective 2-month study. 37 (7.9 ± 3.7 years) had HIV only, and 5 (7.3 ± 2.4 years) also had clinical stroke, all right-sided.

Results: TAMMV were mainly normal in the middle (MCA; right median 96; range 47–147 cm/s, left 100; 64–160) and anterior (ACA; right 82; 46–148, left 80; 35–160) cerebral arteries in those without stroke. TAMMV was higher in the posterior cerebral arteries (PCA) in those with stroke (right 70; 52–157, left 86; 25–154) than in those without stroke (right 56; 33–83, left 56; 33–100) ($p = 0.03$, $p = 0.1$ for right and left respectively). Although left MCA TAMMV was high in one with stroke (median 99; range 54–200 cm/s), there was no difference from those without stroke ($p = 0.9$). In 26 patients with HIV without stroke, CD4% correlated with TAMMV in the left MCA ($r = 0.52$, $p = 0.007$) and ACA; $r = 0.45$, $p = 0.02$) and in the right PCA ($r = 0.48$, $p = 0.01$).

Conclusions: In contrast to adults, children with HIV do not have lower intracranial TAMMV. The increase in PCA TAMMV may reflect compensatory increase in flow in patients with anterior circulation disease. The difference in the relationship between left

and right TAMMV and immune function should be further explored in HIV.

1. *Stroke* 1999;**30**:811–13.
2. *Ann Neurol* 2004;**55**:840–4.

G186 CASE SERIES: CENTRAL NERVOUS SYSTEM COMPLICATIONS OF EAR NOSE THROAT INFECTIONS

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Aims: Highlight CNS complications of ENT infections and its management.
Methods: Descriptive study of cases over the last 12 months (November 2007–October 2008) referred to the neurosciences department.
Results: 10 cases; five males (age 8–14 years, mean age 11 years) and five females (3–14 years, mean age 8 years). Five cases had sinusitis and five cases had mastoiditis as primary ENT infection. They had a combination of CNS infections; eight patients with cerebral and/or subdural abscess, three patients with macroscopic evidence of osteomyelitis, nine with venous/venous-sinus thrombosis and three patients with evident cerebral infarction. Common clinical presentation was headache, high temperature, vomiting, lethargy, agitation, altered sensorium, focal neurological deficits and focal seizures. Common causative organisms for abscess were *Staphylococcus aureus*, Gram-negative cocci and mixed anaerobes. Other organisms were *Strep. milari* and *Pneumococcus*. In addition to emergency and supportive care specific management included: (1) drainage of ENT and/or CNS abscess; (2) broad-spectrum antibiotics, 6 weeks IV and 2–6 weeks oral; (3) steroids for cerebral oedema. Other treatments for complications included: anticoagulation for venous thrombosis for 3–6 months, seizure management. Multidisciplinary team support with neuro-rehabilitation was the key from the beginning. Four of 10 patients had significant morbidity.
Conclusions: CNS complications from ENT infections are not uncommon and outcome is variable. It is our impression that there are increasing numbers of children presenting to neurosciences services with CNS complications of ENT infections. Confirmation of this will involve multicentric study. There are number of possible reasons for this increase including less frequent use of antibiotics in primary and secondary care for ENT infections.

G187 AN AUDIT OF RESPIRATORY CARE IN DUCHENNE MUSCULAR DYSTROPHY

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Background: Patients with Duchenne muscular dystrophy (DMD) develop respiratory failure during their teens. However, there are no definitive UK guidelines outlining respiratory management. Furthermore, a lack of service provision may limit respiratory review. We aimed to compare our management of DMD with published guidelines by the American Thoracic Society (ATS) to determine whether we met their standards.
Methods: A medical student undertook an audit of the medical records of 47 patients with DMD, aged 5–35 years (median 16.5) against standards in the ATS guidelines.
Results: All patients were reviewed regularly ($\leqslant$6 monthly), but forced vital capacity was only recorded on 63% of visits. The proportion of non-ambulant patients who had sleep studies rose from 11% in 2001 to 50% in 2007. 10 of 17 patients who had started non-invasive ventilation (NIV) had done so as an emergency, including one patient who had CO_2 narcosis during a sleep study. Elective initiation mostly arose in patients over 20 years. Ten patients, all of whom were receiving NIV, had cough assist devices and all began after initiation of NIV. Only six of 17 patients who had had spinal surgery had a formal respiratory evaluation before surgery. 21 (45%) patients had Pneumovax documented as being given. Only two patients had documented emergency care plans. Annual chest *x*-rays, blood counts and bicarbonate levels were rare.
Conclusions : The frequency of recorded respiratory assessments in patients with DMD, including lung function and sleep studies, does not meet ATS guidelines. A more frequent respiratory review may allow earlier intervention and avoid emergency care. We have developed a proforma to help achieve this and aim to re-audit. Collection of similar data in a national database may help encourage respiratory surveillance and provide information to support service provision and the development of UK guidelines.

G188 THE IMPACT OF CEREBRAL INJURY ON AMPLITUDE-INTEGRATED EEG MATURATION IN PRETERM INFANTS UNDER 30 WEEKS GESTATION

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Introduction: The impact of cerebral injury on the amplitude-integrated (a)EEG maturation in preterm infants has yet to be defined.
Aims: To quantify the change in aEEG measures during the first and subsequent weeks of life, and to assess the impact of cerebral injury on maturation of aEEG measures.
Methods: Infants born <30 weeks received a median 74 h of continuous two-channel EEG with aEEG monitoring in the first week of life. Infants were classified in the abnormal (AB) outcome group if they died in the neonatal period and/or had grade 3–4 intraventricular haemorrhage and/or moderate or severe abnormalities on cerebral MRI. Four-hour seizure-free epochs of aEEG was analysed for median values of lower and upper margin amplitude as well as the percentage of time the lower margin spends below a threshold of 5 μV.
Results: Of 51 infants recruited, 17 (33%) infants had AB outcomes. For infants with normal or mildly abnormal (NMA) outcomes, the lower aEEG margin ($r = 0.36$, $p = 0.03$) and the percentage of time spent below 5 μV ($r = -0.36$, $p = 0.03$) correlated with increasing gestation for the first recording. Infants in this group showed a significant rise in the lower, mean and upper aEEG margin during the first ($p<0.001$) and subsequent weeks of life. In comparison, infants with AB outcomes had significantly lower aEEG measures that persisted even after adjusting for gestation ($p<0.001$).
Implications: The rise in aEEG amplitudes in the first week of life may reflect ex-utero adaptation as well as maturation in the preterm infant. In the presence of cerebral injury, normal aEEG maturation is disturbed and the aEEG measures may provide a biomarker to select preterm infants for neuroprotective therapies in the future.

G189 LONG-TERM VENTILATION IN CHILDREN WITH SEVERE NEUROLOGICAL DISORDERS

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Introduction: Increasing numbers of children with severe neurological disorders are being ventilated long term. The decision to ventilate is often complicated by medical, ethical and legal dilemmas. The RCPCH guidance suggests five situations where it may be ethical and legal to consider withholding life-sustaining treatment. For children with severe chronic neurological conditions the ''no purpose'' situation can be particularly relevant. In this situation the child may be able to survive with treatment but the degree of physical or mental impairment will be so great that it is

unreasonable to expect them to bear it. Such impairment is present when there is limited or no interaction with the surroundings.
Aim: To evaluate the indications and appropriateness for long-term ventilation in children with severe neurological disorders.
Methods: Retrospective review of children with severe neurological conditions who received long-term ventilation in the last 5 years.
Results: 21 such children were identified. Five children had tracheostomy and 16 had ventilation by mask. 18 were ventilated only at night. The diagnoses included neuromuscular conditions such as Duchenne muscular dystrophy (three), Ullrich muscular dystrophy (two), chronic inflammatory demyelinating polyneuropathy (two), congenital muscular dystrophy, congenital myasthenia, myotubular myopathy, congenital myopathy, spinal muscular atrophy II and undiagnosed progressive neuromuscular disease. Other conditions included mitochondrial disease (three), vasculitis, muscle eye brain disease, transverse myelitis, Crouzon's syndrome and undiagnosed progressive ataxia. Most of these are progressive neurodegenerative conditions while some are static. Only one child had impairment severe enough to cause limited interaction with surroundings (inconsistent smile).
Conclusions: The decision to offer long-term ventilation to children with severe neurological conditions can pose difficult dilemmas. The RCPCH guidance can provide valuable help in this matter. Nearly all children in our group (20 of 21) did not meet the criteria for the consideration of withholding treatment. Ventilation did alleviate respiratory distress and prolong life in this group. It is difficult to assess if ventilation is unbearable in these children.

G190 USE OF EEG IN THE PAEDIATRIC DEPARTMENT OF A DISTRICT GENERAL HOSPITAL

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Aim: Identify if EEG use in a district general hospital is adherent to the National Institute for Clinical Excellence (NICE) guidelines. The diagnosis of epilepsy remains a clinical one. A misdiagnosis of epilepsy has consequences socially, psychologically, pharmacologically and financially for both the patient and family and society. Misdiagnosis rates of epilepsy have been reported to be as high as 30%. Because of this difficulty in diagnosis physicians may put undue emphasis on the results of the EEG. In 2004 NICE produced a document entitled ''Diagnosis and management of the epilepsies in children and adults in primary and secondary care''. This includes guidelines for appropriate use of EEG and in particular it should not be used to exclude or make (in isolation) a diagnosis of epilepsy. It also makes the recommendation that children with suspected seizures should be seen by a paediatrician with an interest in epilepsy.
Methods: Retrospective audit of children referred for EEG over a 12-month period. Demographic data (age, sex), clinician involved, description of clinical findings, reason for EEG, clinical suspicion of epilepsy (low, moderate or high), waiting times, EEG report and final outcome (diagnosis, treatment and medication).
Results: 135 EEGs were performed on 132 patients. EEGs performed on neonates were excluded. Detailed data available on 68 patients with a mean age of 7.4 years (range 1–16 years). The most common presentations were staring/day dreaming (17 of 68) and single generalised tonic clonic seizure (11 of 68). 78% of EEGs were requested by a consultant paediatrician in an outpatient setting. Although only 22% of patients with suspected seizures were seen by a paediatrician with a special interest in neurology/epilepsy. EEGs were requested to ''exclude a diagnosis of epilepsy'' with a low clinical suspicion in 12 of 68 (17.6%) of patients and parental anxiety in two of 68 (3%). In those with a high clinical suspicion of epilepsy, EEG changes supporting the diagnosis were found in 10 of 15 (66%).
Conclusions: To the best of our knowledge there has been limited reporting into EEG use in a district general hospital in children. Our findings support the recommendation of EEG in specific situations (high clinical suspicion of Epilepsy or absence seizures). The requests for EEGs to ''exclude epilepsy'' need to be addressed with local guidelines for EEG use to be implemented and training sessions for all general paediatricians.

G191 TRANSITIONAL CARE: VIEWS OF PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY

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Background: Patients with Duchenne muscular dystrophy (DMD) develop respiratory failure during their mid to late teens, which in the past has been an age when their care is traditionally transferred from paediatric to adult services. We sought to determine the views of patients about transitional care in order to guide service development now that their survival has been prolonged into their twenties and beyond.
Methods: A medical student administered a questionnaire to 40 patients with DMD, aged 10–35 years asking their views on transition.
Results: 22 (55%) patients considered 18–20 years as the appropriate age to move to adult services, nine thought 21–23 years, eight 15–17 years and one gave no answer. Patients were most concerned to know that they would have the same level of care and support and that adult consultants would know as much about DMD as their paediatrician. 37 (92%) patients were unaware of transition clinics—all felt they would be of benefit. Most patients felt that the move to a transition clinic should be 1 year before the move to adult services. Most would prefer the same specialist to be involved in their care from childhood and through adulthood, thus avoiding the need for transition.
Conclusions: Services might consider use of neuromuscular specialists who can provide continuing care for children and young people and through adulthood. Failing that, transition should be started at 17–19 years about 1 year before moving to adult services.

G192 HEADACHES IN CHILDREN, SHOULD YOU BE WORRIED?

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Background: Headaches are common in children and often a cause for worry both for parents and their treating physicians. Though uncommon, brain tumours remain the commonest solid tumour in children. The headache ''red flags in children'' protocol adopted by the American Imaging Management provides guidance for prompt neuroimaging in suspected cases.
Aim: To review the presentation of brain tumours in children and identify features in their history and examination that prompts neuroimaging and enables early diagnosis.
Method: 25 diagnosed cases of brain tumours at the Gloucester Royal Hospital were retrospectively reviewed for signs and symptoms at presentation, complaints to referral and assessment to diagnosis time interval.
Results: Brain tumours were noted in all age groups, including children less than a year and teenagers (12% each). M:F ratio was equal. Supratentorial tumours were as common as infratentorial ones. The majority (68%) were referred by GPs and some via the ophthalmologists. 16% had a 1-day history and the majority (48%) presented within a month of developing symptoms. Headaches, the commonest symptom, was present in 79% (excluding preverbal group) but this was not the sole feature in any of the cases at presentation. Vomiting (60%), visual concerns (56%), balance/co-ordination problems (36%), seizures (24%) and torticollis (16%) were other features noted. The delay in diagnosis occurred as a

result of failure to image possible cases (ignoring long tract signs, unexplained eye signs, seizures difficult to control) or due to the nature of the tumour itself, which required subsequent MRIs for diagnosis.
Conclusions: Headaches per se are not worrying but should be taken seriously if coexistent with another symptom or sign. GPs are most likely to encounter possible cases. Good history-taking skills, complete neurological examination and follow-ups remain the mainstay for early diagnosis. Children may have confounding/unexplained eye signs that warrant early referral to paediatricians.
Recommendations: All health professionals (GPs, health visitors, ophthalmologists and paediatricians) involved with children should be aware of the ''red flag signs'' to refer and investigate possible cases promptly. Following National Institute for Clinical Excellence guidelines for seizure investigation would also help in early diagnosis.

G193 A SIMPLE CASE OF ERB'S PALSY? CASE REPORT

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CP was born by a non-traumatic normal vaginal delivery at term. Initially, he was noted to have a hypotonic left arm and leg; the latter quickly resolved and he was diagnosed with Erb's palsy. Aged 3 weeks, he was referred to hospital, with prolonged jaundice. On examination, he was found to have severe respiratory distress. Managed as possible pneumonia, he rapidly deteriorated requiring continuous positive airways pressure (CPAP) for over 2 weeks. Investigations including FBC, BC, CXR, diaphragmatic and cranial USS were all normal. Inability to wean off CPAP resulted in transfer to regional PICU, where he recovered. On discharge, no cause was found for his respiratory symptoms. Aged 2 months, he was admitted to another hospital, again with respiratory distress. He deteriorated and was transferred to tertiary PICU and ventilated for 36 h. During this admission, he was again found to have a left hemiparesis as well as some new right-sided weakness. An MRI head showed bright right parietal and posterior temporal lobes from inflammation of white and grey matter. After a multitude of other investigations, an EMG and nerve and muscle biopsies were performed showing patch reduction in axonal density and evidence of a demyelinating process and a diagnosis of chronic inflammatory demyelinating polyneuropathy (CIDP) was made. He recovered with 5 days IV immunoglobulins. However, aged 3½ months he attended A&E at local hospital with constipation. Following a suppository, a cyanotic episode with respiratory distress was precipitated, requiring immediate intubation, ventilation and transfer back to PICU. At present (4 months later) he is still on CPAP via tracheostomy. Immunoglobulins, plasmapheresis and methylprednisolone have been little help. CIDP is almost unheard of in babies, and is a chronic form of Guillain–Barré syndrome. It has an incidence of 0.5 per 100 000 in children, but there are few case reports in babies this young. Its heterogeneous presentation makes it easily missed, though it classically presents with numbness, burning pain, progressive weakness and areflexia for more than 8 weeks and has a relapsing and remitting course. Randomised controlled trials show evidence for treatment with IV immunoglobulins and plasmapheresis. 70% make a good recovery, but the relapse rate is high in young people.

G194 CLINICAL PHENOTYPE OF PAROXYSMAL EXTREME PAIN DISORDER, ABNORMAL MAGNETIC RESONANCE IMAGING AND FAILURE TO THRIVE: IS THE DIAGNOSIS SECURE?

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Background: Paroxysmal extreme pain disorder (PEPD) is a rare autosomal dominant condition caused by mutations in the voltage-gated α IX sodium channel (SCN9A) gene. Presentation is seen from the neonatal period with paroxysmal attacks with autonomic features, tonic attacks and syncope triggered by defecation, perineal hygiene or eating. We present an infant with a classical phenotype of PEPD but with contradictory investigation results.
Patient summary: A 4-month-old boy presented with daily paroxysmal episodes since birth, lasting several minutes and consisting of a ''different'' cry, associated with harlequin facial skin erythema, unilateral ptosis, pupillary changes and other autonomic features. The events initially appeared unprovoked but by age 7 months they were triggered by putting objects in the mouth. Video footage of the paroxysmal episodes will be shown. At age 15 months developmental progress is normal and he has no interictal neurological deficit. The infant developed severe eczema and failure to thrive from 6 months.
Investigations: Ictal recording at 4 months revealed no EEG change and repeat at 12 months captured several typical events with no EEG correlate. Brain MRI demonstrated wide sylvian fissures and thickening of the perisylvian cortical grey matter. Metabolic tests were negative. Blood tests for coeliac disease and jejunal biopsy were negative. SCN9A mutation analysis is in progress.
Treatment: Carbamazepine (30 mg/kg per day) has significantly decreased the frequency and severity of events, with increase on drug withdrawal.
Discussion: This patient has the classic phenotype of PEPD, but the MRI findings would usually be seen in a child with anterior opercular syndrome. Failure to thrive further confounds the clinical picture. SCN9A is not expressed in mouse cerebral cortex and cortical dysplasia is not seen in PEPD. A positive SCN9A mutation result would support to the clinical diagnosis of PEPD but on present knowledge would not explain the brain MRI findings.

G195 INTERESTING CASE OF ''PSEUDOTUMOUR''

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A 14-year-old girl presented to us with a history of limping on the right side following a fall down a staircase 6 days ago and landing on her buttocks. She walked somewhat funny initially, which the family put down to an ankle sprain. She was noted later to have a foot drop and loss of sensations below the right knee. Initial neurological examination showed weakness from L4 to S1 level on the right side with corresponding sensory deficit. Left lower limb and upper limb neurology were normal. Deep tendon reflexes were equivocal on both sides and plantar reflexes were flexor bilaterally. She had mild spinal tenderness at T12–L1 level with no deformity or restriction of movement. She also started to have nocturnal enuresis during her stay in hospital. There was also a central disc prolapse at L4/L5 level for which she was conservatively managed with monitoring of neurological status. During a further stay 6 days after her admission she was found to have extensor plantar response on the right side with brisk knee jerk, suggesting a possible upper motor neurone lesion; therefore, she had a MRI scan of head without contrast (as patient refused), which revealed a possible tumour-like lesion involving her left parietal lobe crossing the midline. She was then transferred to neurosurgeons at a tertiary centre for further management. A repeat MRI brain with contrast at the tertiary centre showed a large solitary left parafalcine parietal lobe and splenium of corpus callosum lesion with moderate surrounding vasogenic oedema. The differential diagnosis was between a primary high-grade tumour such as glioblastoma, astrocytmoa, PNET or a plaque of demyelinating necrosis. It was decided to treat conservatively with steroids. The repeat MRI scans showed that the lesion was regressing in size with steroids and hence conservative management was continued. The lesion was

noted to be regressing in size, and no new demyelinating lesions were noted. Her lower limb neurological examination has remained normal. We would like to share our experience and this case highlights that some lesions that look like a malignant lesion on MRI scan may be a benign demyelinating lesion and thereby the importance of giving a guarded prognosis.

G196 NEUROFIBROMATOSIS TYPE 1: LOCAL COUNTY EXPERIENCE

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Neurofibromatosis type 1 (NF1) is an autosomal dominant, multisystem disorder with a myriad of clinical manifestations. The incidence is 2–3 per 2500 and is characterised by its progressive nature and variability of signs and symptoms. Café-au-lait macules, neurofibromas, intertriginous freckling, Lisch nodules and learning disabilities are frequent. Optic and other gliomas, malignant peripheral nerve sheath tumours, and characteristic osseous lesions are also known to occur. This review documents the local experience of the common clinical characteristics of NF1. We also describe the clinical details of patients with uncommon signs and symptoms and considered unusual at presentation. We report on a follow-up of 21 children with NF1. Case notes were screened for age at presentation and documented signs and symptoms. The most common presenting feature was café au lait macules in 20 children (95%). 11 children were diagnosed in infancy. The same number had first-degree relatives with the condition. 13 children had some form of neurodevelopmental problem, mainly speech and language. Four children appeared to be the only people in the family with the condition. Others had a family member, most commonly the mother, with NF1. Four parents received a diagnosis of NF1 following their child's diagnosis. One child presented with hypertension, due to renal artery stenosis. Two children had tumours—one had optic pathway glioma and another had cerebellar astrocytoma. Although the guidelines do not recommend routine scanning in asymptomatic children the diagnosis of astrocytoma needing intracranial surgery have raised doubts in the minds of many clinicians regarding routine imaging. A long-term prospective study looking specifically at asymptomatic individuals of NF1 is needed. We hope that this study will raise awareness about the myriad clinical manifestations in NF1.

1. **Ferner RE,** Huson SM, Thomas N. Guidelines for the diagnosis and management of individuals with neurofibromatosis. *J Med Genet* 2007;**44**:81–8.