NEUROLOGY

G218 CAUDATE AND HIPPOCAMPAL VOLUMES, AND BEHAVIOURAL AND COGNITIVE PERFORMANCE OF VERY PRETERM INFANTS IN EARLY ADOLESCENCE

Abernethy L, Palaniappan M, Cooke RWI *Department of Radiology, and Department of Child Health, University of Liverpool at Royal Liverpool Children's Hospital (Alder Hey), Liverpool L12 2AP, UK*

The caudate nuclei and the hippocampus are both involved in the functions of short-term memory and learning. The latter is particularly susceptible to damage from stress, and has been shown to be altered in size in epilepsy, Alzheimer's disease, schizophrenia and post-traumatic stress disorder. Preterm infants (PT) often show learning, motor and behavioural difficulties at school. This study aimed to determine whether structural differences shown on magnetic resonance imaging (MRI) in these areas of the brain were associated with these deficits in PT in adolescence.

94 adolescents (86 PT, 8 term) who had been assessed at school at 12-13 years for motor impairment (Movement ABC), attention deficit and hyperactivity disorder (ADHD, Child and Adolescent Psyciatric Assessment), and intelligence (IQ, Wechsler Intelligence Scale for Children III), underwent MRI of the brain at 15-17 years. Volume measurements of the caudate nuclei and hippocampus were made bilaterally for each child, and the ratio of the left to right structure calculated as a percentage. The neuroradiologist was blind to the results of earlier testing.

Caudate and hippocampal volumes and the hippocampal ratio were significantly higher in the term controls. No significant relationship of volumes or ratios to motor impairment was observed. PT with ADHD had a significantly smaller right and left hippocampus. PT with a low IQ (<85), had a significantly smaller right caudate nucleus and left hippocampus, and a lower hippocampal ratio when compared to those PT with a higher IQ. Standardising for sex and handedness using logistic regression did not alter these findings.

These volumetric differences suggest that specific structural changes during development of the brain in PT may be responsible for behavioural and learning deficits in later life.

G219 SEVERE HYPERKINETIC MOVEMENT DISORDER IN A CHILD TREATED WITH DEEP BRAIN STIMULATION: A CASE REPORT

Gill S, Curran AL, Tripp J, Malerikos L, O'Sullivan K, Stanley O *Bristol Children's Hospital, St Michael's Hill, Bristol*

Introduction: Deep stimulation of brain structures has been in use for adult movement disorders since 1987 {Pollack, 1997}. Its use in childhood movement disorder has not previously been reported in the literature . We would like to report its use in one such case.

Clinical history: This eleven year old male is the first child of unrelated parents. Foetal movements were reduced in the third trimester. Born at term by normal delivery. He was noted to be hypotonic in the neonatal period. Athetoid movements developed in the first year of life. Epilepsy with tonic/clonic and partial motor seizures occurred at 18 mths. He crawled at 3 yrs, and walked with aids at 5 yrs. He developed a multi-focal, stimular sensitive and action myoclonus with an exaggerated startle reflex which was non-progressive at 5 yrs. Detailed investigations have all been normal. Intellectual function is good. Medical management of his movement disorder has included carbamazepine, sodium valproate, haloperidol, tetrabenazine, benzhexole, acetazolamide and phenytoin. A trial of clobazam precipitated status choreaticus with severe rhabdomyolysis complicated by hyperpyrexia, renal impairment and disseminated intravascular coagulation.

The failure of medical treatment led to him being referred for assessment for neurosurgical intervention. Following detailed evaluation, he underwent insertion of deep brain stimulators (Medtronic Itrel II IPG) into the posteroventral globus pallidus bilaterally.

Results: The patient's movement disorder has shown sustained improvement at 9 months follow-up. The family is now able to socialise and take trips in a way that was previously impossible. Orthopaedic intervention is now being planned as consistent, supported weight bearing by the patient is now possible. He is no longer injuring himself frequently and is starting to gain weight. Gross Motor Score, 'posting', and other evaluations have been performed with the stimulators in the 'on' and 'off' state. These results will be available within the next 6 weeks.

G220 CHANGING TRENDS IN THE INVESTIGATION AND MANAGEMENT OF FEBRILE CONVULSIONS IN THE FIRST YEAR OF LIFE: LOCAL EXPERIENCE AND A REVIEW OF THE LITERATURE

Carroll WD *Department of Paediatrics, City General Hospital, Stoke-on-Trent*

The UK guidelines state that lumbar puncture should 'almost certainly' be performed following a first febrile convulsion in the first year of life. A retrospective audit was carried out to determine if practice has changed over the last ten years at our centre.

Methods: Patients were identified using the hospital admissions books for 1989 and 1998. The notes of all those between 6 months' and 1 year with an admitting diagnosis of convulsion, fit or seizure were examined individually. For those who satisfied the diagnostic criteria for a febrile convulsion data were retrieved to determine the patient characteristics, seizure duration, investigations done, subsequent management and final diagnosis.

Results: 30 children between 6 months to a year of age were admitted to our centre following their first febrile convulsion (17 in 1998 and 13 in 1989). The demographics of both groups were very similar. There was a significant decrease in the number of children undergoing lumbar puncture (0/17 in 1998 vs. 8/13 in 1989 ($p<0.01$)) and a trend towards less blood tests, less antibiotic use and shorter hospital stays in 1998.

Discussion: Systematic review of the literature demonstrates that it is extremely rare for bacterial meningitis to present as a febrile convulsion. In the absence of other physical signs (drowsiness, bulging fontanelle or rash) then the maximum incidence of bacterial meningitis is approximately 0.1% (6/5532). Furthermore, the diagnostic yield of lumbar puncture in this group is only 66% (4/6) which raises questions about the value and risks of lumbar puncture in this age group.

G221 LUMBAR PUNCTURE FOLLOWING THE FIRST FEBRILE CONVULSION: A SURVEY OF PRACTICE IN THE UNITED KINGDOM IN THE FIRST YEAR OF LIFE

Carroll WD *Department of Paediatrics, City General Hospital, Stoke-on-Trent*

Guidelines produced by the British Paediatric Association state that paediatricians should 'almost certainly' perform a lumbar puncture in children under one year, even in the absence of signs of meningitis as these may be occult. A survey of 100 paediatric registrars was performed to determine current practice across the UK.

Methods: 120 hospitals were randomly selected from the Directory of Hospital Telephone Numbers. From these, 100 paediatric registrars from hospitals in every region in the UK were contacted by telephone. Firstly they were asked if there were any guidelines concerning the management of febrile convulsions in their department. Then they were asked whether they would perform a lumbar puncture on a ten-month-old child who had had its first fifteen-minute febrile convulsion. They were all told that the child had a runny nose a temperature of 38C but no abnormal physical signs (bulging fontanelle, drowsiness or residual focal neurology).

Results: 46 (out of 100) registrars said that they would and 54 said that they would not perform a lumbar puncture. The presence or knowledge of departmental guidelines had little effect on the likelihood of performing a lumbar puncture.

Local guidelines	Lumbar puncture
Yes 42	Yes 21 (50%)
	No 21 (50%)
No 28	Yes 11 (39%)
	No 17 (61%)
Don't know 30	Yes 14 (47%)
	No 16 (53%)

Conclusion: There is currently no consensus amongst middle grade paediatricians on whether lumbar puncture should be performed following a first febrile convulsion in the first year of life.

G222 NORMAL DECUSSATION OF CORTICOSPINAL AXONS AND AXONAL PROJECTION TO LUMBAR SPINAL SEGMENTS IN HUMAN SUBJECTS WITH L1CAM MUTATIONS

Dobson C, Villagra F, Clowry GJ, Kenwrick S, Donnai D, Miller S, Eyre JA *Developmental Neuroscience, Department of Child Health, University of Newcastle upon Tyne NE1 4LP*

Background: L1CAM gene (Xq28) mutations are linked to neurological syndromes characterised by spastic diplegia. The L1CAM knockout mouse shows failure of corticospinal (CST) axonal decussation and projection beyond the cervical spinal cord. Our hypothesis was that similar pathology underlies the spasticity of males hemizygous for L1CAM mutations.

Methods: Studies were performed on 7 adult carrier females and 9 hemizygous males (aged 2–29 years). Transcranial magnetic stimulation (TMS) excited the CST and responses were recorded bilaterally in biceps brachii, abductor digitii minimii and quadriceps femoris. Subthreshold CST conditioning of the stretch reflex of biceps and quadriceps was performed. The CST was examined in post-mortem brain and spinal cord material from a one month old male with L1CAM mutation.

Results: TMS evoked contralateral and ipsilateral responses in all three muscles, which were in the normal range in upper limb muscles but had abnormally high thresholds and delayed onsets in quadriceps. Ipsilateral responses were smaller, of higher threshold and delayed relative to contralateral,indicative of CST decussation. The patterns of CST modulation of the stretch reflex in biceps and quadriceps were abnormal, suggesting a reduced

projection to inhibitory interneurones at both segmental levels. Abnormality of CST function was greater in hemizygous males than in carrier female. Post mortem examination of the CST revealed normal decussation and axonal projections to lumbar spinal segments.

Conclusions: These data show abnormalities of CST function in hemizygous males and ''carrier'' females which is greater in the projection to the lower limb. However, anatomical and neurophysiological evidence reveal CST axonal decussation and projection to the lumbar spinal segments. While these results support the hypothesis that L1CAM has an important role in CST development, they do not indicate significant abnormality of CST axonal guidance as suggested by the L1CAM knockout mouse.

G223 SUCCESS RATE AND SAFETY OF ORAL SEDATION

Ahmed SR, Robson J *Darlington Memorial Hospital, Hollyhurst Road, Darlington*

Background and aims: A child's co-operation is needed for a number of procedures and investigations. Paediatricians commonly use oral sedation. Anaesthetists strongly discourage the use of heavy sedation in children and advocate general anaesthesia as an alternative and safe option. However waiting time for general anaesthesia is often too long and the facility is not available in small district general hospitals. The Aim of this study was to find safety and success rate of oral sedation.

Methods: A combination of Trimeprazine (2mg/kg) and Triclofos (30mg/kg) was given approximately 90 minutes before the planned procedure/investigations. Fasting was per hospital's policy for general anaesthesia. Exclusion criteria were raised intracranial pressure, potential airway obstruction/respiratory failure, risk of aspiration of gastric contents and severe renal or hepatic failure. A nurse trained in Advanced Paediatric Life Support supervised all procedures. All children had their saturation and vital signs monitored throughout the procedure. Indications for sedation were variable and included MR scan (43%), ERA (18.3%), MCUG (17.8%), EEG (9.9%), CT scan 5.5% and others 5.5%. Success was defined as deep or conscious sedation and completion of intended investigation or procedure.

Results: Over a 2 year study 202 sedations were performed in 178 children. Mean age of children recruited in the study was 3 years and 2 months. Overall success rate was 90%. 77% children were sedated in less than 90 minutes and 88% completely recovered within 4 hours. No complication of sedation was recorded and all children were able to go home the same day.

Conclusion: Oral sedation is a safe and practical option where a young child's co-operation is required for a procedure or investigation. Careful selection and monitoring however, is essential.

G224 AN AUDIT OF CHILDHOOD EPILEPSY MANAGEMENT AT A REGIONAL PAEDIATRIC NEUROLOGY SERVICE

Barklay A, Richardson G, Curran AL, Gardner C, Sharples PM *Bristol Children's Hospital*

Introduction: Recent literature has highlighted the importance of audit in the management of childhood epilepsy[1-3]. National standards of care were approved by the BPNA,1996. Using these guidlines, an audit of epilepsy management at a tertiary paediatric neurology clinic was conducted.

Aim: To audit management and outcome after one year of all newly diagnosed children with epilepsy.

Method: A retrospective case note study using a data collection form was performed on all children who had a first referral for an electroencephalogram to the neurophysiology department Nov 1997-Nov 1998.

Results: Of 329 new referrals for EEG, 325 case notes were reviewed. Of these 71 were diagnosed as epileptic. 51% were male and 49% female. Compared to the BPNA standards our results were as follows (target is 100% in each case): all children should have a specific diagnosis by 1 year (85% - 11% had a change in diagnosis during the year); should have documented advice regarding epilepsy associations (1%); where indicated should see a paediatric neurologist (100%); where indicated should have an MRI scan (88%); should receive either carbamazipine or sodium valproate as first line treatment (except for specific indications e.g. West's Syndrome) (100%); should have a documented reason for change in medication (83%); should have documentation of their development and education (77%); should have documentation of physical examination (69%); should only have sodium valproate levels measured to establish compliance (100%).

Conclusion: Our service scored >80% on 7/10 standards. Documentation requires improvement scoring <80% in 3/10 standards. Further work will include a proforma in the notes to be completed at each outpatient visit which will facilitate re-audit.

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G225 INHERITED EARLY ONSET SEVERE AXONAL POLYNEUROPATHY WITH RESPIRATORY FAILURE AND AUTONOMIC INVOLVEMENT

Mohan RU, Misra VP, Britto J, Muntoni F, King R, Thomas PK *Paediatric Intensive Care Unit, St Mary's Hospital, London*

We report dizygotic twin infants with early onset of severe axonal polyneuropathy presenting with respiratory failure due to diaphragm weakness. A female sibling who had had been similarly affected died from respiratory failure at nine months of age. Both twins had persistent tachycardia, increased sweating and hypertension secondary to autonomic involvement. This is an unreported presentation of presumed autosomal recessively inherited hereditary motor sensory neuropathy.

Twin male black infants born at 38 weeks gestation presented at six months of age with acute respiratory failure requiring mechanical ventilation for over six weeks following which they underwent tracheostomies to receive continuous ventilatory support. There was no maternal polyhydramnios or parental consanguinity. Neurological examination revealed generalised hypotonia, wasting and flexion contractures of distal limb muscles with absent deep tendon reflexes. Cognitive function was normal. Bilateral diaphragmatic paralysis was confirmed by ultrasound. Persistent hypertension refractory to betablocker therapy responded to phenoxybenzamine. Extensive investigations on serum and urine, imaging studies of brain, kidney and heart were normal. EMG showed profuse fibrillations at rest. Mixed median nerve action potentials and sensory action potentials were absent. Nerve biopsy showed reduced proportion of myelinated axons with preservation of unmyelinated axon population and axonal degeneration. Muscle biopsy revealed typical features of denervation. Duplication of chromosome 17p11.2 was not detected in either infant. The twins are now 10 months of age and continue to receive ventilatory support.

Congenital forms of axonal polyneuropathy causing early respiratory failure are extremely rare. To conclude, this description would seem to be a unique disorder, the gene defect for which is unknown.

G226 MRI APPEARANCE AND CLINICAL SIGNIFICANCE OF PUNCTATE LESIONS IN THE NEONATAL BRAIN

Cornette L, Toh V, Ramenghi L, Tanner SF, Arthur RJ, Childs AM, Martinez D, Ridgeway JP and Levene MI *Leeds General Infirmary, Department of Paediatrics, Clarendon Wing, Leeds*

Aims: Although considered as having a haemorrhagic origin, the clinical significance of punctate lesions in neonatal brain MRI is largely unknown. Our aims were to study the incidence of these lesions in a large cohort of newborns and to relate anatomical characteristics to clinical outcome.

Methods: Punctate lesions were characterised for *number* [<3, 3-10, >10]; *appearance* [clump, linear]; *distribution* [anterior, middle, posterior; uni- or bilateral]; *signal intensity (SI)* [on T_1, T_2]; *association* with minor [cortical highlighting, discrete oedema] and major [malformation, PVL, infarction, intraventricular bleeding, cortical atrophy] abnormalities.

Results: We analysed 110 MR scans (92 neonates, born in 1998). Punctate lesions were observed in *15/50 preterm neonates*. The number of lesions were either <3 (n=3, all clumps), 3-10 (n=5, 3 linear) or >10 (n=7, 6 linear). SI was uniformly increased on T_1 and reduced on T_2. The lesions were seen periventricularly and in the centrum semiovale (14/15), posterior (8/15) and anterior (3/15). All other brain abnormalities were related to asphyxia and were either minor (11/15) or major (4/15). In preterms without punctate lesions (35/50), brain abnormalities were either absent (18/35), minor (4/35) or major (13/35). Punctate lesions were seen in *2/42 term neonates*, organised in clumps, with only minor additional brain abnormalities. Clinical outcome of infants with isolated punctate lesions (i.e. involving only minor additional brain abnormalities) is highly favourable (n=13, mean age=14.5 months, delayed development of language in 1 patient).

Conclusions: Punctate lesions are predominantly seen in preterm neonates, most commonly linearly organised and bordering the lateral ventricles, their origin most likely being haemorrhagic. Caution is needed in interpreting *isolated* punctate lesions as being pathologic, since so far most of these patients have a normal developmental outcome.

G227 AN ATYPICAL CASE OF ALPERS' SYNDROME

Chow G, Johnston DI, Mellor D, Smith J, Jaspan T *Queen's Medical Centre, Nottingham*

Case Report: We present a case of a boy aged 1 year 7 months with mild developmental delay and poor growth. Both this boy and his elder sister were on the ''at risk register''.

Investigations showed elevated serum and CSF lactates with generalised aminoaciduria and abnormal liver function tests. Neurophysiology, neuroimaging, and skeletal survey were normal. Skin fibroblast cultures showed normal fatty acid β oxidation studies, pyruvate dehydrogenase and pyruvate carboxylase, and muscle biopsy was normal on histochemistry and EM. Cytochrome oxidase IV was low normal.

He was admitted at 1 year 10 months to ITU with left sided convulsive movements. He was floppy with a reduced level of consciousness. EEG

showed focal electrical status epilepticus over the right hemisphere. There was worsening of liver function tests and clotting results. A CT scan was normal, however an MRI scan 2 days later showed diffuse cortical thickening, hyperintensity and bilateral thalamic lesions on T2 weighted images becoming more pronounced on a follow up MRI scan. CT scanning by this stage demonstrated global cerebral swelling and hypodensity and a typical "cerebral reversal sign". Despite ventilation and treatment with anticonvulsants he continued to have myoclonus and died of a respiratory tract infection. Postmortem findings were obliteration of occipital lobe medially, and sponginess involving frontal and parietal lobes. The liver showed fatty change, bile duct proliferation, inflammation and nodular regeneration.

Conclusion: This was an atypical case of Alpers' disease as intractable seizures occurred very late after his initial presentation. His later MRI scans were consistent with severe hypoxic ischaemic encephalopathy.

G228 GUILLAIN-BARRE SYNDROME AND JAPANESE ENCEPHALITIS PRESENTING AS ACUTE FLACCID PARALYSIS IN SOUTHERN VIETNAM

M Gainsborough[1], VC Kanh[2], T Solomon[1], NM Dung[2], R Kneen[1], B Wills[1], TTN Thuy[2], SR Smith[3], HJ Willison[4], JJ Farrar[1] *[1]Wellcome Trust Clinical Research Unit and [2]Centre for Tropical Diseases, Cho Quan Hospital, Ho Chi Minh City, Vietnam [3]Dept of Clinical Neurophysiology, National Hospital for Neurology, London [4]University Dept of Neurology, Southern General Hospital, Glasgow*

As part of the survillance programme for poliomyelitis, all children with an acute flaccid paralysis (AFP) admitted to a infectious diseases hospital in southern Vietnam were studied prospectively between 1996–1999.

15 and of 43 children were diagnosed with Guillain-Barre syndrome (GBS) clinically and 9 patients had evidence of acute Japanese encephalitis (JE) infection. Compared with the JE cases, the GBS cases were more likely to have had a preceding illness, arm involvement, symmetrical and distal weakness, whereas the JE cases were more likely to have fever at onset of weakness, neck stiffness, urinary retention and the later development of encephalopathic features. 3 GBS and 2 JE patients developed respiratory failure and one patient died in each group. All surviving GBS patients made a functional recovery by 12 months but 7 with JE had severe residual flaccid weakness resembling the sequelae of paralytic poliomyelitis.

Electrodiagnostic criteria divided the GBS cases into 3 with acute inflammatory demyelinating polyneuropathy (AIDP), 11 with acute motor axonal neuropathy (AMAN) and 1 unclassifiable. The AMAN group only differed clinically from the AIDP group in having no sensory deficits and less bladder involvement. However 7 patients in the AMAN group had raised levels of antiganglioside antibodies compared to none of the AIDP cases, and 4 of these had anti-GD1a antibodies, a feature also highly significant in cases of AMAN reported in north China.

As the incidence of polio declines in Vietnam, knowledge of the predominant local causes of AFP needs to be revised. Distinguishing GBS and JE myelitis, may be difficult based on admission features. However making a specific diagnosis is important because of the different prognosis and for planning future preventitive measures.

G229 MOYAMOYA DISEASE IN A NORTH-EASTERN POPULATION

Dabydeen, L Forsyth, RJ Ramesh, V *Department of Paediatric Neurology, Newcastle General Hospital*

Moyamoya disease is a rare cerebral vasculopathy of unknown origin and has primary and secondary types. The primary form is most commonly seen in Japan where it affects nearly 4000 people.

Aims: to document the presentation, features and natural history of Moyamoya within a Caucasian population.

Methods: retrospective case notes review of all children with Moyamoya under our care

Results: 6 children were identified with primary Moyamoya disease. 3 presented with stroke, 1 with learning disabilities and migraine, 1 with transient ischaemic attacks and 1 with migraine and choreoathetosis. Age at presentation ranged between 9 months (stroke) and 8 years (choreoathetosis). Apart from the 2 children who presented directly to our unit with stroke where a diagnosis of Moyamoya was confirmed in the acute period, this took between 3 11 years in the remaining group. All 4 children presenting under the age of 4 years had moderate learning disabilities. 3 of the children had revascularisation procedures. 1 child had surgery within 1 month of presentation and has a normal cognitive profile; the other 2 children had their surgery more than 5 years from presentation and have learning disabilities. Of the children presenting under the age of 4 years, 2 are siblings with an affected mother, the third has an affected mother and the fourth child's mother has not yet been imaged.

Conclusion: Familial Moyamoya occurs in 10% of the affected Japanese population and has recently been localised to Chromosome 3p. Although Moyamoya occurs too rarely in Caucasians to be able to get a clear idea of inheritance patterns, in our very small series we found that half the children had an affected parent. Young children at presentation had a poor prognosis.With the advent of MRA , screening of first degree relatives should be considered.

G230 THE DEVELOPMENT OF A CONSENSUS BASED GUIDELINE FOR THE MANAGEMENT OF THE CHILD WHO HAS HAD A SEIZURE

Armon K*, Stephenson TJ*, MacFaul R**, Eccleston P*, Werneke U″, Smith S+, Williams L++. **Child Health, University of Nottingham. **Pinderfields General Hospital, ″London School of Hygiene and Tropical Medicine. +Paediatric and ++A&E Departments, Queens Medical Centre, Nottingham*

Introduction: Seizures affect 3% of children and account for 5% of all medical attendances to paediatric accident and emergency (A&E).

Aim: To develop an evidence and consensus based guideline for the child who presents to A&E having had a siezure, using the Delphi method.

Method: A systematic search of electronic databases 'Medline', 'Cochrane' and 'Embase' was performed. Relevant quality publications were graded according to their strength of evidence. Statements were made with a graded level of recommendation. Publications used, the literature review and derived statements were sent to a national panel (selected to be broadly representative) of 30 medical and nursing staff. They were asked to rate their level of agreement with each statement on a 1-9 Likert scale and to comment. Consensus agreement was pre-defined as 83% of panelists rating 7-9. The results were fed back in two rounds to the panel with amendments for further rating and comments.

Results: 42 statements were made, 2 were based on level III evidence, 7 implied by level III, 23 on level V and 10 were derived from panellists comments. 10 reached consensus in round 1, 14 in round 2, 10 in round 3 and 8 did not achieve consensus. These 8 were alternatives to those that did, or concerned issues that could be decided at a local level.

Conclusions: This process resulted in a comprehensive evidence and consensus based guideline on the management of the child presenting to A&E having had a seizure. An analysis of the impact of this guideline will be presented separately.

G231 WORSTER-DROUGHT SYNDROME (W.D.S.) A MILD TETRAPLEGIC PERISYLVIAN CEREBRAL PALSY: A REVIEW OF 47 CASES

Clark M, Neville BG, Carr L *Great Ormond Street Hospital For Children NHS Trust and Institute of Child Health (UCL) London*

Aims: To clarify the diagnosis of Worster-Drought Syndrome and to record associated features and complications.

Methods: Retrospective case-note analysis of 47 children thought to have W.D.S. on clinical grounds i.e. a congenital upper motor neurone bulbar palsy.

Results: The study children had *significant bulbar problems* (with 80% still needing a modified diet and a similar number with speech impairments requiring augmentative communication methods at last review). There were also high rates of *predictable bulbar complications* (86% had dribbling: 60% had glue ear: gastro-oesophageal reflux in 40%; history of poor nutrition in 40%; aspiration in 40%;). Most of the children had *additional complex impairments* (91% had mild pyramidal tetraplegia; 81% learning impairments; 60% malformations/deformations; 41% neuropsychiatric problems and 28% epilepsy). Over half of the children had significant medical problems in the first year but the mean age at diagnosis was 6 years.

There were no obvious causes in pregnancy or birth. 6 children had a family history of W.D.S. 32% (12/37) had abnormal neuroimaging including 5 with bilateral perisylvian polymicrogyria.

Conclusion: In our experience W.D.S. is not uncommon, is relatively easily diagnosed and is crucial not to miss as the management of these children's multiple impairments is complex and requires a careful team approach.

W.D.S. is clearly a form of cerebral palsy, as a syndrome that includes motor impairment arising from static damage to the brain in early life. Its core elements are a suprabulbar paresis, a mild spastic tetraplegia and a significant excess of cognitive and behavioural impairments and epilepsy.

G232 MAGNETIC RESONANCE IMAGING OF THE NEONATAL CENTRAL NERVOUS SYSTEM ON THE NEONATAL INTENSIVE CARE UNIT

Whitby EH, Paley MN, Smith MF*, Sprigg A, Yong W*, Griffiths PD *Academic Department of Radiology, University of Sheffield and Neonatal Intensive Care Unit, Jessop Hospital, Sheffield**

Aims: To assess the role of a dedicated magnetic resonance scanner on a neonatal intensive care unit.

Methods: A 0.2 Tesla permanent magnet system (Niche, Innervision) was installed in a room adjacent to the NICU. Imaging protocols were optimised on 60 normal controls. Subsequently MR imaging was performed on 40 babies with suspected intracranial pathology (32 had pathology). All patients had spin echo T1 weighted images, 25 had spin echo T2 weighted images and 3 had diffusion weighted imaging. All images were reported independently by two experienced radiologists. The results were compared with the clinical, cranial ultrasound report, which was performed within 24 hours of the MR scan.

Results: One hundred babies have been successfully scanned to date, 68 normal and 32 with pathology. Of the 32 cases with pathology the ultrasound and MR results were in agreement in 13cases. In 16 the MR images provided

additional information to the ultrasound and this had management implications in 7 patients. Two patients had normal ultrasounds and the MR showed middle cerebral artery infarcts. One patient had a large posterior fossa bleed and another had schizencephaly not seen on ultrasound. One had severe hydrocephalus on ultrasound, which was proven as hydranencephaly with MR imaging. Hypoxic Ischaemic changes were seen in 2 patients several days earlier on MR than Ultrasound. In 3 cases ultrasound demonstrated grade one intraventricular haemorrhages not seen on the MR images.

Conclusion: Low field strength MR imaging provides a valuable method of imaging and provides additional information to ultrasound.

G233 COLLODION BABIES WITH HYDROCEPHALUS AND LARGE HEAD, A NEW SYNDROME?

Sinha GP *Manor Hospital, Walsall WS2 9PS*

Ichthyosis has been noted in several syndromes, ie Sjögren Larsson syndrome, (global delay, epilepsy, diplegia), Refsum syndrome (ataxia, neuropathy, etc), Rud syndrome (epilepsy and mental impairment) and Tay syndrome.

However we are unaware about its association with hydrocephalus. We present two brothers with ichthyosis born to non consanguineous white parents.

Case one, now a three year old, was noted to have parchment like skin, ectropion, cracking of skin at knee and groin and contractures of knee, ankle and elbow. His weight and head circumference were between 10th and 50th Centile.

His skin seems to be improving with treatment. Unfortunately at 6 months of age he presented with vomiting and had clinical signs of hydrocephalus. Cranial CT scan had confirmed that hydrocephalus was due to acqueduct stenosis. He has required revision of shunts since then. He is displaying delay in all areas of development and is being treated for epilepsy. We would show clinical photograph, growth chart and cranial CT scan.

Case two, now a one year old, had parchment like skin and low set deformed ears. His skin is much better. His developmental milestones are normal. Now he has a rather large head (weight between 50th-75th Centile and head circumference 75th-90th Centile) but so far has not developed hydrocephalus.

We have excluded X-linked ichthyosis due to steroid sulfatase deficiency and feel that ichthyosis is of autosomal recessive origin. We would highlight the significance of measuring head circumference and monitoring development in children with ichthyosis. If our second case develops neurological problems then we would have to expand our battery of investigations.

G234 CLINICAL AND ELECTROENCEPHALOGRAPHIC SEIZURES IN COMA: RELATIONSHIP TO OUTCOME

Kirkham FJ[1], Wairui C[2], Newton CRJC[1,2] *[1]Neurosciences Unit, Institute of Child Health (University College, London), [2]KEMRI Unit, Kilifi, Kenya*

Aims: Prolonged and multiple seizures are associated with brain damage in humans. Status epilepticus may occur in acute encephalopathy but prognosis has been assumed to be related to aetiology and the severity of underlying process rather than to the seizures. This study examines the hypothesis that the prognosis for coma in childhood is determined, at least in part, by the presence of seizures and/or status epilepticus.

Methods: The patients were observed by members of the nursing and medical teams and any clinical signs suggestive of seizures were noted. A cerebral function analysing monitor (Medaid) was used to monitor either 1 or 2 channels of EEG. The presence, number and duration of any EEG seizure discharges in an individual patient were noted and were compared with survival and outcome (normal or moderate handicap=good; severe handicap, vegetative state or death=poor) using the Mann-Whitney U-test.

Results: 188 patients (median age 2, range 0-15 y; 52% male) with a wide range of aetiologies (113 hypoxic-ischaemic encephalopathy, 45 cerebral malaria, 11 encephalitis, 2 meningitis, 7 head injury, 3 Reye's syndrome and 7 miscellaneous) were studied. Survival was related to aetiology (p=0.0001), Glasgow coma score on admission (p=0.0001), EEG score on admission (p=0.0001) and mean arterial pressure on admission (p=0.004), but not to number (p=0.3), duration (p=0.5) and duration of the longest seizure (p=0.7). Outcome was related to aetiology (p=0.0001), Glasgow coma score on admission (p=0.0001), EEG score on admission (p=0.0001), mean arterial pressure on admission (p=0.007), and to number (p=0.006), duration (p=0.004) and duration of the longest seizure (p=0.003).

Conclusions: Although survival and outcome for coma in childhood may be predicted at least in part on admission, seizures after admission appear to be a potentially modifiable cause of neurological morbidity and warrant interventions designed for prevention or treatment.

G235 MUTATIONS IN THE TRKA GENE IN KUWAITI PATIENTS WITH CONGENITAL INSENSITIVITY TO PAIN WITH ANHYDROSIS (CIPA)

A Moosa[1], M Baloushi[2], E Ismail[3], S Mardy[4], Y Muira[4], Y Indo[4] *Departments of Paediatrics, Kuwait University[1], Mubarak Al-Kabir[2] & Farwaniya[3] Hospitals Kuwait and Kumamoto University School of Medicine, Japan[4]*

We describe mutations in the TRKA gene in 7 children from 5 consanguineous Kuwaiti families with CIPA. CIPA is a rare autosomal recessive disorder characterise by unexplained fevers, anhydrosis, mental retardation and absence of pain resulting in self-mutilation of fingers, tongue, lips and joints. The TRKA gene encodes a tyrosine kinase receptor for nerve growth factor, which ensures survival of embryonic sensory & sympathetic neurones. The following mutations were found: A single base deletion in exon 1 (c.284 del A.) of the gene in 2 sibs and a G to A substitution in the 5' splice site of intron 7 (IVS7 + IG>A) in another 2 sibs (These have been reported in Am.J.Hum.Genet.64: 1570, 1999).

Two unrelated children had a novel C to T transition at nucleotide 610 (c.610 C>T) in exon 5, which changes Gln to a termination codon at amino acid 176 (Gln 176X). The parents of all these 6 patients were heterozygous. The last patient showed an 8 base-pair insertion in exon 13, but the mother did not show the same mutation. Mutations of the TRKA gene are associated with CIPA.

G236 THE IMPACT OF IDIOPATHIC GAIT DISORDERS

Wassmer E, Wright E, Rideout S, Whitehouse W *Neurology & Physiotherapy Department, Children's Hospital, Birmingham*

Many children are in hospital with gait disorders requiring treatment, some physical, some idiopathic. The prevalence of idiopathic gait disorders in children is unknown. The impact of school absence, investigations and therapy may be substantial. The intent of this study was to estimate the incidence and impact of idiopathic gait disorders in children admitted with gait disorders in a tertiary children's hospital.

We evaluated prospectively all the children admitted with a gait disorder requiring treatment in the form of physiotherapy at Birmingham Children's Hospital, using a standardized pro forma, during a three-month period (March and June 99).

103 children (ages 2 to 16) were admitted with gait disorders (57 female). 45 children were admitted under orthopaedics, 32 neurosurgery, 26 other. Eight (ages 10 to 15) had an idiopathic gait disorder (5 female). All 8 children exhibited function impairment, pain and school absence. Some had lethargy (6) and sensory symptoms (4). Most reported an antecedent illness (5) and a similar illness history in a acquaintance (4). All children were above average at school and most had many friends (7). Results were normal of physical and laboratory and imaging examinations.

The symptoms resolved after therapeutic admission (1 week to 6 months).

Idiopathic acquired gait disorders are 7.7% of gait disorders in admitted children. The economic and social impact is substantial with regard to diagnosis, investigations, treatment and school absence.