A CLINICAL EVALUATION OF VISION ASSESSMENT IN CHILDREN WITH SENSORI-NEURAL DEAFNESS

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Since 1991, children with sensori-neural deafness and their families in the Southern Derbyshire Health Authority have been supported by a holistic multi-agency approach to their diagnosis and management.

Analysis of 122 children assessed at the Child Development Centre has shown a high number with additional vision problems. The ages of the children ranged from 8 months to 16 years, and 72 had newly diagnosed sensori-neural deafness, the majority under 5 years. Of these children, 66 had congenital sensori-neural deafness associated in 15 with specific syndromes, 3 being related to vision. Eleven had acquired deafness, and there were 45 of unknown cause. The severity of deafness varied from mild to severe, and also 6 cases of Usher syndrome, mainly type 2. Some children had more than one eye defect.

The reported findings are that deaf and hearing impaired children are three times more likely to develop ocular abnormalities than their hearing peers, which makes early detection of paramount importance. An important outcome has been the establishment of local guidelines for both vision assessment and screening for Usher syndrome by electro-physiological testing at aged 7 years and above. Screening has been completed to date in 71 children with congenital sensori-neural deafness. More research is needed to address ethical issues of screening the children ranging from 8 months to 16 years, and 72 had newly diagnosed sensori-neural deafness, the majority under 5 years. Of these children, 66 had congenital sensori-neural deafness associated in 15 with specific syndromes, 3 being related to vision. Eleven had acquired deafness, and there were 45 of unknown cause. The severity of deafness varied from mild to severe, and also 6 cases of Usher syndrome, mainly type 2. Some children had more than one eye defect.

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TELOMERE TESTING IN CHILDREN WITH SEVERE LEARNING DIFFICULTIES

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Background and aims: Recent advances in cytogenetic techniques to detect submicroscopic chromosome rearrangements involving telomeres have refocused attention on children with severe learning difficulties of unknown aetiology. The aim of our study was to investigate the usefulness of telomere analysis in such children.

Methods: A casenote review of the 83 children attending Stanley school for children with severe learning difficulties on the Wirral was carried out and those with no diagnosis were ascertained. Previous investigations in these children, including karyotype, were negative. Those with accompanying dysmorphic features and/or a positive family history of chromosomal anomalies were selected. Blood samples were taken from these for telomere analysis using the Vysis Total Vision probe kit.

Results: 16 of the children (19%) attending the school had no diagnosis. 9 of these (11%) fitted our criteria for telomere analysis. Consent for blood testing was obtained from 8 of this group. Fluorescence in situ hybridisation studies using telomere probes for all the autosomes and X chromosome were carried out and revealed no evidence of deletion or rearrangement of any of the telomeric regions in this group.

Conclusions: Despite applying selective criteria no positive results were obtained from this small sample. However the importance of a diagnosis for families and thus more accurate genetic counseling should not be underestimated and needs to be balanced against the cost (£150 per test) and the low yield from this test.

A PROSPECTIVE, POPULATION BASED ASSESSMENT OF DEVELOPMENTAL DELAY AND CRANIOFACIAL DYSMORPHISM IN CHILDREN BORN TO MOTHERS WITH EPILEPSY

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Children of women with epilepsy are at increased risk of developmental delay, craniofacial dysmorphism & malformation.

Aims: 1) To determine the true incidence of developmental delay in our prospectively recruited, non-hospital based cohort of children aged 2—3 years. 2) To identify the aetiological risk factors involved. 3) To establish whether delay occurs independently of malformation and craniofacial dysmorphism.

Methods: One hundred and eleven children have been assessed. The Bayley’s scales are used to assess Mental Developmental Index (MDI). Facial measurements are taken with calipers and clinical photographs obtained.

Results: Of 111 children, 8 had normal development (79%) and 25 delayed performance (21%), (p = 0.0215). The proportion with severe delay (MDI > 2 SDs below mean), (8/111 = 7.2%) is substantially greater than the background rate of 1.5%, and is highly statistically significant (p = 0.006). We found a significant relationship between developmental delay and poor maternal educational attainment, (p = 0.0118). There also appears to be a significant correlation between developmental delay & in utero exposure to CBZ (p = 0.0340), and VPA polytherapy (p = 0.0049). Seizures in pregnancy are unrelated to developmental delay (p = 0.453). We have found no correlation between dysmorphism, malformation & developmental delay. This is irrespective of drug dose.

Conclusions: Children born to women with epilepsy are at significantly increased risk of developmental delay. This may be associated with maternal intelligence & certain anticonvulsants. It occurs independently of malformation & craniofacial dysmorphism.

ARE WE STILL LATE IN PICKING UP DUCHENNE MUSCULAR DYSTROPHY?

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Objective: To identify the time interval between presentation of symptoms and confirmation of diagnosis of Duchenne muscular dystrophy (DMD).

Design: Retrospective review of hospital medical records of children diagnosed with DMD.

Setting: Walsall Health District of West Midlands, with population of 60,000 children.

Method: All cases of DMD currently being cared for in Walsall were identified, and records reviewed.

Results: There were 7 boys with the diagnosis of DMD. (1) Two of the cases were identified after DMD was diagnosed in siblings. (2) Confirmation of DMD in two siblings led to the termination of an affected foetus. (3) The average of presentation was 28.4 months (6m to 49m). Presenting symptoms were waddling gait (n=2), frequent falls (n=1), speech delay (n=1), difficulty in climbing stairs (n=1). In 3 cases limited abduction of hips in early infancy may be an early presentation of DMD. (3) None presented with late walking (over 18 months). (4) In 3 cases limited abduction of hips in early infancy led to hospital referral. However this did not directly lead to the suspicion and diagnosis of DMD. (5) Average age of confirmation of diagnosis was 47.6 months (12m to 68m). (6) Average delay in the confirmation of diagnosis was 19.2 months (6 months to 28 months).

Conclusion and remarks: (1) There is great diversity in the presenting symptoms of DMD. (2) Limited abduction of the hips in early infancy may be an early presentation of DMD. (3) None presented with late walking (over 18 months). (4) There continues to be a significant delay between the onset of symptoms and the confirmation of diagnosis of DMD.

COGNITIVE IMPAIRMENT IN 6–8 YEAR OLD CHILDREN FOLLOWING NEONATAL ENCEPHALOPATHY

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Aims: Few studies have evaluated outcome for children without disability following neonatal encephalopathy (NE). We have performed a regionally-based study of outcome in middle childhood for such a group. NE was defined as moderate or severe encephalopathy thought to be due to hypoxia before birth after 35 weeks gestation.

Methods: Children born between 1992–94 were identified from the Trent regional database and examined at school with a matched classmate as a control, using the British Ability Scales 2e and NEPSY.

Results: 45 cases have been evaluated between 78 and 95 months of age. Of these, 71% were attending special schools and were not matched to controls; 6 had cerebral palsy (4 quadriplegia) and 3 had multiple disabilities. Scores for the remaining children are shown in the table.

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In this population there was a gradation of performance across the 3 groups. The ventilated NE group consistently scored lower compared to controls.

Conclusions: Children who survive moderate or severe encephalopathy with or without disability are at risk of later neuro-cognitive dysfunction and outcome appears to relate to the severity of neonatal condition.

Audit of an ADHD Clinic with Particular Reference to the Use of Risperidone for Associated Oppositional Defiant Disorder and Conduct Disorder

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Audit of a clinic population regarding diagnosis and management, particularly 104 children with associated Oppositional Defiant Disorder (ODD)/Conduct Disorder (CD). Additionally the clinic’s use of risperidone was analysed. The children attended a multidisciplinary clinic with a national referral basis, specialising in neuropsychological and learning problems, especially ADHD and related conditions.

64% had combined ADHD and of these 57% had associated early onset ODD and/or CD. 73% of this latter group were not adequately treated by Ritalin or Dexedrine alone, requiring a second medication—generally initially clonidine—to achieve a 92% very positive outcome. 30 children aged 6–21 years were treated with risperidone, usually because of persistent ODD/CD. Interval between initial diagnosis and institution of risperidone treatment, between 0 months and 6 years. Dosage: between 0.5mg and 6mg daily. 50% required a bd dosage. Risperidone treatment duration in these patients—up to 6 years. 25 (67%) showed moderate or very significant improvement. In 5, risperidone was stopped because of no improvement (2) or excessive weight gain (3). Other side effects: vomiting, drowsiness or withdrawal dyskinesia (1). Liver function tests were normal.

Preliminary data suggest significant improvement can be achieved in children with ADHD and comorbid early onset ODD/CD by more intensive management, often with more than one medication. Risperidone may have a significant place in the management of these children where other treatments have proved unsuccessful.