Accuracy of clinical diagnosis in Down’s syndrome

Hindley and Medakkar\(^1\) showed that the clinical diagnosis of Down's syndrome is inaccurate in one third of cases. We can imagine how stressful it will be for the parents if they have been told that their child may have Down's syndrome and then subsequently the karyotype proves to be normal. We conducted a retrospective study to estimate the accuracy of clinically suspicion in our region and in our hospital in particular.

Using the regional cytogenetic laboratory database, all clinically suspected cases of Down's syndrome born in the West Midlands region during the period June 2000 to December 2002 were identified and karyotype results analysed. All babies identified from Birmingham Women's Hospital were studied in detail by reviewing the case notes. Of 233 suspected cases from the whole West Midlands region, 148 cases were positive by karyotype. Hence the accuracy of clinical suspicion was 64%. These figures were similar to results from Hindley and Medakkar,\(^2\) which showed this was 68% nationally and 64% in the Manchester region. However, from Birmingham Women's Hospital, of 29 cases identified, 25 had a karyotype of trisomy 21 and so a higher accuracy rate of 86%.

We cross checked the patient data from Birmingham Women's Hospital with the rest of the region and found that there were no missed cases from our hospital. Based on the information given to parents before doing the karyotype, in 22 babies where parents were told the diagnosis of Down’s syndrome was felt to be certain, karyotype was positive in all 22. However, in seven cases where they were told a positive diagnosis was possible, four had a normal karyotype. All 25 cases that had a normal karyotype were all under three days of life; in two of the babies who were preterm, it took at least 3–4 weeks for clinical suspicion to develop. When we analysed the four negative cases, two were tested without being assessed by a consultant. One case was tested just based on profound hypotonia at 31 weeks but no other clinical features. In the final case, karyotyping was done to reassure the parents because there was reported suspicion by two independent midwives and a registrar, but the consultant felt the baby was normal.

Our data from Birmingham Women’s Hospital showed a favourable accuracy rate compared with the previous study.\(^1\) This can be explained by the fact that the tertiary hospital may have more experienced neonatologists compared to the broad cohort of junior and senior paediatricians involved in other parts of the region. We believe that assessment by a senior paediatrician before testing may minimise the risk of negative results. There may be difficulty in diagnosing Down's syndrome in preterm babies who may take some time to manifest classic features. We also agree with Hindley and Medakkar\(^1\) that some sort of scoring system like Fried’s index\(^3\) may also be useful in improving the accuracy of clinical diagnosis. However, a large and prospective study is needed to evaluate those scoring systems.

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References


Beware CSF pressure measured under general anaesthesia

Wraige et al\(^2\) describe three children suffering from idiopathic intracranial hypertension (IIH) in the absence of papilloedema.\(^3\) MRI findings in two cases along with an initial symptomatic improvement following lumbar puncture support the diagnosis. In the third case MRI scan was normal, and the child’s headaches did not result to lumbar puncture or acetazolamide. In all three cases CSF pressure was measured under general anaesthesia with control of position and of carbon dioxide concentration, presumably by end-tidal CO2 monitoring. The anaesthetic technique is not reported.

The message that children with IIH may not have papilloedema is a valuable one. However, we would like to add a note of caution regarding the measurement of CSF pressure under general anaesthesia. We have found unexpectedly raised CSF pressure when performing lumbar punctures under sevoflurane anaesthesia, administered to facilitate MRI scanning. In children with a variety of neurological disorders.

All inhalational anaesthetic agents have a cerebral vasodilating action and will increase cerebral blood volume and hence intracranial pressure (ICP). In addition the spontaneously breathing child will sustain an appreciable rise in ICP from the respiratory depressant action of these drugs and consequent hypercarbia. This increase can be prevented by controlled ventilation. Volatile agents may also reduce cerebral perfusion pressure by their hypotensive effect which is due to a combination of systemic vasodilatation and direct myocardial depression; in higher doses cerebral autoregulation may be abolished altogether.\(^4\)

Obstruction to venous return also increases ICP. The flexed position of the anaesthetised child during lumbar puncture may be more marked than when performed without anaesthesia. Coughing and straining at induction of anaesthesia and obstruction to respiration from bronchospasm or the introduction of positive end expiratory pressure (PEEP) will all cause a rise in ICP which may remain a factor at the time of lumbar puncture. Finally, end-tidal CO2 is always lower than arterial CO2 and it is not always possible or desirable to check a blood gas prior to lumbar puncture.

It seems likely that the use of general anaesthesia to facilitate lumbar puncture will increase as deep sedation on paediatric wards becomes less acceptable and increasingly skilled paediatricians perform fewer lumbar punctures. We are concerned that children having lumbar puncture under general anaesthesia could be erroneously diagnosed as having intracranial hypertension. Until a standard anaesthetic technique is developed which can be shown to have a minimal effect on intracranial pressure, we believe that measurements of CSF opening pressure under general anaesthesia should be interpreted with caution. If doubt exists, and certainly if surgical treatment is contemplated, insertion of an intracerebral transducer allows definitive measurement of ICP over a period of hours or days.

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References


Cat scratch disease presenting as meningomyeloradiculopathy

We report meningomyeloradiculitis as a presenting picture in a child with cat scratch disease (CSD) and identify the need to include this infection in the differential diagnosis of meningomyeloradiculopathies. We also show the likely benefit of antibiotic therapy.

Case report

An 11 year old girl presented three months prior to hospitalisation with low back pain radiating to the lower limbs with paresthesia, limp, and a progressively decreasing strength in the right lower limb. She had sphincter disturbances with frequency of micturition and dysuria. A month later, she developed a stiff spine with impairment in daily activities. Initial examination (day 0; D0) showed decreased strength in the right lower limb with thigh atrophy and reduced sensations. There was scoliosis (dextroconvexity). MRI (fig 1) revealed enlarged conus medullaris, with hypertetraesthesia on T2. Cerebrospinal fluid (CSF; sub-occipital puncture) showed lymphocytic meningitis.
Serologic immunofluorescence revealed a Bartonella henselae (BH) IgG titre of 1/1024; IgM was 1/128. Interrogation revealed cat exposure. Ofloxacin 30 mg/kg/day and rifampicin 20 mg/kg/day were given for six weeks. Recovery in general state and usual activities started a few days after treatment initiation. Improvement of scoliosis followed. All motor, sphincter, and sensory disturbances gradually recovered over one month. On D60, full spine x-ray, T1 and T2 weighted spinal cord MRI, and CSF normalised. BH serology showed a negative IgM, and IgG was at 1/512. At the last assessment (D90), clinical examination was normal.

Comment
CSD is a self limited infection. Neurological involvement is rare; full recovery in these cases has been reported but may take several months. In these neurological forms, antibiotics were suggested to accelerate resolution. In our patient, combined antibiotic therapy resulted in dramatic improvement, supporting such a therapeutic approach.

The spectrum of infiltrative or space occupying lesions of the central nervous system (CNS) is wide and includes infectious/parasitic conditions (such as mycobacterial infection, angiostrongylosis, and schistosomiasis), inflammatory, vascular, and metabolic diseases (“pseudotumoral” acute disseminated encephalomyelitis, sarcoidosis, Langerhans histiocytosis, GM2 gangliosidosis, and venous infarcts). CSD related CNS infective/inflammatory conditions have therefore to be added to this spectrum of infiltrative pathologies and should be carefully excluded before resorting to an invasive technique such as CNS biopsy.

References

Intra-renal reflux
Intra-renal reflux may accompany high-grade vesico-ureteral reflux (VUR) and represents the severe end of the VUR spectrum. In addition, intra-renal reflux is usually seen in very young patients. Presence of intra-renal reflux is a high risk factor for renal scarring, which is an important cause of chronic renal failure and arterial hypertension in children.1 When Angulo et al investigated VUR, they documented intra-renal reflux in 17/89 kidney units in 61 patients with VUR.2

Voiding cystourethrography remains the gold standard for the diagnosis of VUR1 and is one of the best modalities to demonstrate intra-renal reflux, if present. This is often seen as a wedge or fan shaped flux of contrast starting from the calices outlining the renal papillae, and may extend to the surface of the kidney (see fig 1).

Early recognition of VUR and prompt management favourably influences the prognosis3 and hence all children at risk should be screened.4 In particular, children with intra-renal reflux should be considered for early intervention to stop reflux (either by endoscopic correction or ureteric implantation) and have regular follow up to monitor renal growth and renal function.

Only wholeness leads to clarity
Authors Lee and Mann argue for law compelling use of cycle helmets by children to prevent road deaths and serious injuries.1 This observer is surprised that the peer reviewers allowed publication of material lacking evidence either that the actual risks faced by child cyclists justify compulsion, or that the real world results of helmet compulsion in other countries justify compulsion in this country. These shortcomings are typical of papers in the medical literature that attempt to address the issue of cyclist safety. I believe that these chronic shortcomings are primarily the consequence of the failure of the peer review process.

In the first place, it is irrational that consideration of helmet laws for children is restricted only to cycling, or even begins with cycling. Although, tragically, around 30 child cyclists have been killed on public roads annually in recent years, typically 110 child pedestrians are killed annually.2 Estimates of death risk per kilometre travelled derived from standard data sources3 do not suggest that child cyclists face greater risks than child pedestrians in most age groups. It is in any case evident that the average child is almost four times more likely to be killed in the casuality while walking rather than cycling. The peer reviewers ought to have insisted on a more general discussion of the risks faced by children in transport. This would have placed the injuries to cyclists in context and enabled priority, surely the basis of any systematic approach to public health interventions.

In the second place the evidence for the effectiveness of cycle helmets is split by an interesting contradiction. The authors cite research based on case-control trials reporting that helmeted cyclists were much less prone to serious head injuries than the bareheaded, at least at the time and in the locality of the research work. However, there is also a substantial body of evidence based on population-level studies of head injuries with increasing helmet use. These studies consistently fail to show material benefit for cyclist populations that took up helmet wearing. This was even true in New Zealand, where cyclists responded willingly to helmet promotion, with voluntary use climbing 60% even before the well obeyed law of January 1994 came into force.4 The famous helmet laws for the states of

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The factoid that child cyclists are more at risk from motor traffic than child pedestrians and that cycle helmets can protect children in road traffic accidents. A famous line from Schiller comes to mind as apt to the occasion:

Nur die Fuelle fuehrt zur Klarheit.

[Only wholeness leads to clarity]

Let us hope, for the sake of the public understanding of cycling, that in future peer reviewers apply this wisdom. There will be resurgence of children walking and cycling only when the perceived danger from motor traffic in urban areas is addressed. Proposing compulsory use of inappropriate safety equipment evades this simple truth. Public health interventions must focus on the source of the perceived danger, not burden the innocent with the consequences of adult licentiousness.

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References


BOOK REVIEW

Pediatric gastroenterology and clinical nutrition


This book, although a paperback, is quite substantial, weighing in at 1.25 kg on my scale. It is notable for being the first of its kind in this field. The authors are from the UK, and the book is written in English.

In summary, the book is a valuable addition to the field of pediatric gastroenterology. It is well-written, clear, and comprehensive. It is highly recommended for anyone working in this field, and it is a must-have for any library.

R A F Bell

PROOFREADER

This book covers the major aspects of gastroenterology and includes sections on pancreatic and liver disease; there are also valuable sections on eating disorders and food aversion.

This book is just the job wanting to go a bit beyond the standard texts, such as “Nelson” and “Forfar and Arneil” and its fairly large print makes it easy to read for those, such as MRCPCH candidates, reading chapter by chapter, and those of bifocal age, whereas the rather poor index, for which the large font is a disadvantage, does not help its use as a quick reference; there was no mention, for example, of probiotics in the index, yet Lactobacillus rhamnosus GG and Lactobacillus casei are mentioned in the treatment of acute diarrhoea and inflammatory bowel disease respectively.

The enormous amount of space (179 pages) devoted to appendices rather unbalances the book for the cursory reader, although the information contained therein could be a godsend for someone needing to prescribe special dietary supplements, or to understand a dietician’s advice, such as a paediatrician with significant numbers of children with gastroenterological disorders.

The discrepancy between its excellent crisp chapters of text and the bulky reference section makes me wonder just at whom this book is targeted; perhaps a clue to this dichotomy is to be found in the page of acknowledgements, where Dr Lifschitz states: “This work is a publication of the US Agriculture, Agricultural Research Service (USDA/ARS) and the Children’s Nutrition Research Centre... It has been funded by the USDA/ARS under cooperative agreement No. 6250–51000.” That may explain why, despite two of the three authors being from London, the text is in American English: this really isn’t a problem since the differences between diarrhoea and diarrhoea and coeliac and celiac are slight.

If the appendices and an improved index could be printed in smaller text, this would be an even better, yet less bulky, book.

Archivist: John Snow’s theory of rickets (Arch Dis Child 2004;89:147). In this article the composition of alum is incorrectly given as potassium aluminium phosphate. Alum actually contains sulphate, not phosphate. The error is much regretted.

In the Arch Dis Child supplement I of this year (published in April) the author details of abstracts GB3 (pA32) were not published. They are as follows: J. Hart, C. Harrison, C. Andersen, for The Mercy Neonatal Nosocomial Infection Working Group. Department of Paediatrics, Mercy Hospital for Women, East Melbourne, Victoria.