Accuracy of clinical diagnosis in Down’s syndrome

Hindley and Medakkar 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 karyotype proves to be normal. We conducted a retrospective study to estimate the accuracy of clinically suspicious 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 Midland 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 Midland 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,

The accuracy of clinical suspicion was 64%. These figures were similar to results from Hindley and Medakkar, but showed that the use of general anaesthesia for lumbar puncture is undesirable to check a blood gas prior to lumbar puncture.

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 were confirmed positive were seen by a consultant before testing. In 23 of 25 babies, clinical hypotonia occurred within the first 2 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 clinical suspicion to develop. When we analysed the four negative cases, two were tested without clinical suspicion to develop. 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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 antibiotherapy.

Case report

An 11 year old girl presented three months after initial diagnosis of severe depression, and a progressively decreasing strength in the right lower limb. She had spasticity of both lower limbs, with frequent micturition and dysuria. A month later, she developed a stiff spine with impairment in all lower limb movements. Initial examination (day 1) showed decreased strength in the right lower limb with reduced sensation. There was scoliosis (dextroconvexity). MRI (fig 1) revealed enlarged conus medullaris, with hypointensity 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 DBS, 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

CSF is a sell 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 infections/parasitic conditions (such as mycobacterial infection, angiostrongylosis, and schistosomiasis), inflammatory, vascular, and metabolic diseases (“psychomotoral” acute disseminated encephalomyelitis, sarcoidosis, Langerhans histiocytosis, GM2 gangliosidosis, and venous infaracts).11 CSF 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 patients with VUR.2

Voiding cysto-urethrography remains the gold standard for the diagnosis of VUR and is one of the best modalities to demonstrate intra-renal reflux, if present. This is often seen as a wedge or fan shaped flush of contrast starting from the calyces 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.

Figure 1 Arrows show the presence of intra-renal reflux.

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.5 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.6 Estimates of death risk per kilometre travelled derived from standard data sources7 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 become a serious injury or pedestrian than a cyclist (see table 1). 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 reaching 60% even before the well obeyed law of January 1994 came into force.1 The famous helmet laws for the states of...
Australia brought into effect during 1990–94 drew a similar null result on close analysis.1 In the United States, population-level data gathered by the Consumer Product Safety Commission (a US government organisation that strongly promotes helmet use) shows that the risk of head injury per US cyclist increased by 40% during the 1990s, while helmet use increased from under 20% to at least 50% of cyclists.2 The omission of such evidence places a serious question mark against the competence of the peer reviewers in this case.

The hiatus between clinical trials and population-level results is of scientific interest and draws the curiosity of inquiring minds. Ignoring the hiatus smoothes the existence of a mystery. This is unscientific.

It must be added by-the-by that studies of reported casualties in Britain have revealed a disturbing tendency towards increasing severity of injury with increasing helmet use. This has been observed at the national level and for London,3 where helmet use grew much earlier than the national average. Edinburgh has been identified as having the highest level of helmet use in the country. An ongoing analysis of reported casualties by the author has revealed increasingly severe injuries after 1995, especially for child cyclists. These increases cannot be accounted for by worsening road conditions, since this would have been revealed in pedestrian injury trends. It is not absolutely clear whether the effect is coincidence or consequence, but fair peer review ought to have insisted on some commentary on whether helmet use has influenced reported road casualties. The objective is, after all, to reduce deaths and serious injuries in road crashes.

That helmet use has failed to improve reported road casualties is not surprising. A cycle helmet is designed to meet the event of a simple fall at speeds below 12 mph. Such a mild crash is unlikely to incur serious injury when road riding. Safety campaigners are pressing helmets to an application for which they were not intended. The ethics of this are questionable, a point peer reviewers should have highlighted. The use of helmets is more relevant off-road or “at play”, stunt riding on BMX or MTB type machines. The use of helmets in such situations is perhaps to be encouraged, although parental supervision should come first. On the other hand, these comparatively high risk activities are the consequence of children being denied the freedom to cycle for transport. Riding sensible road bikes on public streets either is, or ought to be, a safe mode of travel for children, not rationally to be distinguished from walking.

In summary, the peer review process has failed to stop incomplete evidence being presented as reliable knowledge. The readership may in consequence be led into two levels of misconception:

- The factoid that child cyclists are more at risk from motor traffic than child pedestrians
- The factoid 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 kitchen scales. Its 963 pages include more than 350 pages of text and references, the rest being devoted to extensive appendices on normal values and recommended dietary intakes for just about everything, and also the contents of many therapeutic foods and supplements. Furthermore, the font size in the text and index (12 lines per 2 inches, compared to 15 in “Nelson”) is rather larger than that usually found in medical texts.

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 plantarum 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 cursive 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.

R A F Bell

Corrections

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 abstract G38 (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.

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Arch Dis Child 2004 89: 692-693
doi: 10.1136/adc.2003.042101

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