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Is the current threshold level for screening for congenital hypothyroidism too high? An audit of the clinical evaluation, confirmatory diagnostic tests and treatment of infants with increased blood spot thyroid-stimulating hormone concentrations identified on newborn blood spot screening in Wales

The UK Newborn Screening Programme has set standards for the identification, investigation and early management of children with congenital hypothyroidism.[1] The standards state that infants with a blood spot thyroid-stimulating hormone (TSH) concentration greater than 20 mU/l are considered to have a positive screening result and referred for investigation by a designated clinician as defined by the British Society for Paediatric Endocrinology and Diabetes. Infants with an initial blood spot TSH concentration between 10 mU/l and 20 mU/l are considered to have a ''borderline'' result, and the assay is repeated. If the concentration remains greater than 10 mU/l on reassay, a repeat blood sample is collected. If the repeat blood spot result is greater than or equal to 10 mU/l this is considered a positive result and the infant referred for further investigation. A blood spot concentration less than 10 mU/l is considered a negative result and not pursued further. In Wales, infants with a blood spot TSH concentration between 5 mU/l and 10 mU/l are also referred for investigation.

The cut-off concentration of greater than 5 mU/l was chosen after the Perkin Elmer autoDELFIA neonatal hTSH assay (Perkin Elmer Life Sciences) was introduced in March 2003. Comparison of the Perkin Elmer assay with the established method (an in-house adaptation of the ACS 180 Bayer ACS TSH method (Bayer)) showed that the Perkin Elmer assay results were approximately 50% lower. A cut-off of 10 mU/l had been used with the in-house assay.

There are 16 screening laboratories in the UK, including Wales. Three use a cut-off concentration of 5 mU/l, four use 6 mU/l, three use 8 mU/l and six use 10 mU/l. Therefore, there is considerable variation in the cut-off screening threshold across the UK despite the screening programme standards.

Between 1 January 2003 and 31 December 2004, 41 infants in Wales were detected with a blood spot TSH concentration greater than 5 mU/l (23 had an initial TSH greater than 20 mU/l, 8 had an initial TSH between 10 mU/l and 20 mu/l and 10 had an initial TSH between 5 mU/l and 10 mu/l). Of the 10 infants with an initial TSH concentration between 5 mU/l and 10 mU/l on the blood spot, 9 had an increased plasma TSH concentration on initial evaluation (mean TSH 20.6 mU/l, range 6–30.1), although this normalised on repeat analysis in 6 infants. Normalisation of TSH occurred between 4 weeks and 3 months.

Three infants with persistently raised TSH concentrations were given thyroxine (age of commencement between 22 days and 31 days). Of these, one has remained off treatment after a trial discontinuation off treatment at the age of 2.5 years (TSH 30.14 mU/l at commencement of treatment) and the other two infants have remained on treatment. One infant has Down syndrome and has remained on relatively small doses of thyroxine (37.5 μg) up to the age of 2.5 years with a maximum TSH concentration of 6.6 mU/l (TSH 80 mU/l at commencement of therapy). The other infant has required increasing doses of thyroxine due to persistently raised TSH concentration (TSH 14.5 mU/l at commencement of therapy; and TSH 13.1 mU/l, free T_4 12.3 pmol/l at 9 months of age) suggesting this to be a permanent form of congenital hypothyroidism.

This audit has identified a case of congenital hypothyroidism that would not have been detected with the recommended standards set by the Newborn Screening Centre. In Wales, infants identified with initial blood spot concentrations between 5 mU/l and 10 mU/l will continue to be investigated and followed up.

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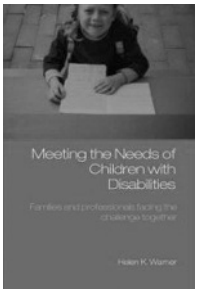
BOOK REVIEWS

Meeting the needs of children with disabilities

Edited by Helen K Warner. Published by Routledge, London, 2005, £22.99 (paperback), pp 176. ISBN 0-415-28038-9



This book addresses areas identified by the English National Board as essential for student nurses. However, the appeal would be wider than this, and includes paediatricians, trainees, therapists and family doctors. Those in education would find some of the chapters very relevant too, and would be heartened by the recognition of the overlap between health needs and education needs in the lives of children and young people with disabilities.

There are a number of very useful and apposite chapters that I have been dipping into over the last months when I should have been writing this review. The first one I looked at, by the editor Helen Warner, was on meeting the fundamental needs of children with disabilities, in this case the needs of such children in hospital. I had thought that my developmental paediatric patients would have had good experiences when admitted to our paediatric hospital, but in fact I had just had feedback of a very unsatisfactory admission. When arranging for the teenager in question to come in for overnight oxymetry, I had failed to mention her intellectual disability. As a result, no bed was made available for a caregiver, and the young person and her parent had to move to another ward late at night so that the caregiver could stay with her. As Dr Warner points out, communication needs to be excellent.

Then, when planning some research into the needs of families of children with autism spectrum disorders, I opened the book again. Guess what. The chapter by Claire Thurgate on ''The importance of respite care'' gave me a quick review of current theory and literature around this crucial issue. The same author has also written a useful chapter on transition planning, putting more emphasis on the educational side of things than is usual in our local practice where health and education are often quite separate.

There are other excellent short chapters, for example on pain management, and on the importance of movement and play. An omission, in my view, is the lack of any discussion of the role of gastrostomy feeding in children with disability and feeding difficulties in the chapter on feeding and eating. In my experiences there are certainly children where gastrostomy feeding has been a very positive move for both the child or young person and their caregivers. I for one would want student nurses and other readers to be aware of this valid option.

I would recommend this book to student nurses, and to people like me who already work with children with disabilities and need updating and reminding of what is important. It would be a good book for paediatricians to