Letters to the editor

'An international perspective on Tourette syndrome'

SIR–The paper by Freeman et al.[1] offered a unique insight into the epidemiology and clinical features of Tourette syndrome (TS). TS is now being redefined from a rare condition to one which is relatively common (perhaps 3% of school children[2]) and one which exists on a spectrum with normality[3]. Co-morbid conditions are also taking a more prominent position. However, there are several instances of data from this study that are markedly at odds with existing data.

The mean prevalence of coprolalia in the studies was 14%: considerably less than many other studies of clinic populations with TS.[4, 5, 6, 7] This also applies to the prevalence of self-injurious behaviours (SIB). This is given as 14% also (compared with other studies estimating between 33% and 48%).[8, 9] The percentage of people in the study with learning difficulties (23%) is also higher than would be expected and may point to a variety of aetiologies for tics (a number of conditions present with both tics and learning difficulties e.g. congenital errors of amino acid metabolism). Also, including those with learning difficulties may start to stretch the diagnostic criteria of TS (i.e. not due to a general medical condition or explained by 'other conditions').

I was also surprised by the relatively low prevalence of obsessive–compulsive behaviour and obsessive–compulsive disorder (OCB/OCD; 27% and 32%). Many other studies have placed the prevalence of these symptoms far higher (up to 90%).[8, 10, 11, 12] Does the presence of OCD in patients in this study mean that they cannot be included in the OCB category?

I am struck by a different pattern shown by these data compared to previous studies. This is a population with a large proportion of people who have learning difficulties, a high proportion with ADHD, and a relatively low level of symptoms which have stronger phenomenological links with TS (coprolalia and OCB/OCD). Does this reflect changing diagnostic trends amongst clinicians, changing referral patterns, a transatlantic difference (69% of patients were from North America) or a genuine change in the syndrome itself? However we view it, TS is being redefined. Will this be beneficial to patients or researchers?

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'Freeman et al. reply'

SIR–We want to thank Dr Rickards for giving us an opportunity to explain some aspects of our paper on Tourette Syndrome (TS) which may have been unclear. The database has now grown in both size and representation, so in the following description we have used a re-analysis of 4132 participants from 61 sites in 25 countries (percentages are given in whole numbers).

It is important to remember that our paper referred to reports from a large number of clinics, each with varying patterns of referral and ascertainment, but all with considerable experience in diagnosing TS. Previously reported studies, to some of which Dr Rickards refers, were usually based upon cases at a single site, and at a time when TS was much less well known.

Dr Rickards suggests that we are 'redefining' TS, but we believe that he is referring only partly to the official definition (which indeed has changed over time), and partly to reported prevalence and to the perception of what is associated with it. These are very different considerations.

The ranges of reported coprolalia and self-injurious behaviour (SIB) vary widely in each geographical region. Because the database began in North America, and the contributions of other countries are now catching up, we have now undertaken a more comprehensive data analysis from the published tripartite method (Canada, the USA, and all other countries), as can be seen in Table I. Table II displays the very wide range of reported prevalence in each region. As presented in our paper, clinics having a high proportion of cases with comorbidity show higher rates of both coprolalia and SIB. It is likely that the difference from the older rates cited are accounted for by this factor.

There is confusion again between 'learning difficulties' and 'learning disability' as used in the UK and traditional usage that continues elsewhere (meaning mental retardation). Dr Rickards refers to 'stretching the diagnostic criteria' by 'including those with learning difficulties'. DSM-IV and the new DSM-IV-TR[1] do refer to organic conditions in which

one should not diagnose TS. But the 'learning difficulties' to which Dr Rickards refers are those vaguely defined problems that the clinician thinks are important (whether or not assessed by a neuropsychologist), and might be due to a combination of factors – but not mental retardation. The rate of mental retardation in the TIC database sample is 3.8%: not out of line with that reported for the general population. Most clinicians working with individuals with cognitive limitations are aware that they may present typical characteristics and symptoms of TS over and above any stereotypies that may also be present.

Dr Rickards is puzzled by the 'low prevalence of OCB/OCD'. Obsessive–Compulsive Disorder (OCD) has an arbitrary requirement of at least 1 hour of symptoms per day, but obsessive–compulsive behaviours (OCB) that do not reach that level may still be clinically significant. (To confuse matters further, in some parts of the world OCD is referred to as OCB.) Since the dividing line is arbitrary and the training and experience of clinicians differ, it is more revealing to combine the prevalence of the two categories. The results are shown in Table II. It can be seen that these reported rates are not very different from those reported previously, nor do they differ very much regionally. Hebebrand and colleagues[2] reported the combined OCB/OCD rates as 33%. As cited in their paper, others have reported different rates. To reply to another question, of course since OCB and OCD are poorly demarcated segments on a continuum, one cannot be said to have both.

Finally, Dr Rickards wonders whether our report reflects changing diagnostic trends (possible), referral patterns (most likely), transatlantic differences (there is no evidence of real differences), 'or a genuine change in the syndrome itself?' We would not be able to determine the latter with certainty. But since the rates of reported SIB and coprolalia in non-comorbid cases do not differ significantly between North America and Europe, we see no reason to conclude anything other than that multiple factors, known and unknown, are involved in determining the characteristics of the referred cases. Our paper and our continuing study aim at showing that, regardless of varying reported rates of comorbidity, there are still clearly identifiable associations. These associations shed light on both the pattern of comorbidities we see in specialty clinics and help to distinguish the problem behaviours frequently encountered clinically from whatever TS itself may turn out to be. In the long run, it seems reasonable that this will be beneficial to all concerned.

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Table I: Distribution of 4132 participants by geographical region (%)

Region	Proportion of database
North America (Canada/USA)	68
Europe (including Turkey)	22
Africa	<1
Asia	3
Australia	2
South America	2
Middle East	2

Table II: Selected items by regions with many cases (%)

Item	All Cases	Europe	N. America	Asia	Mid. East
TS Only	12	13	11	18	14
ADHD	60	56	64	52	55
OCD	25	15	30	24	17
OCB	33	41	31	24	33
OCD+OCB	58	56	61	48	50
Coprolalia[a]	14 (5)	21 (5)	10 (4)	29 (10)	21 (8)
SIB[a]	15 (4)	23 (3)	12 (3)	21 (10)	–

[a]Rates in 'TS Only' cases in parentheses.

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'Non-invasive technique for assessment and management planning of oral–pharyngeal dysphagia in children with cerebral palsy'

SIR–We read the article by Selley and colleagues[1] with interest. The Exeter Dysphagia Technique (EDAT), clearly described in this paper, provides specific and detailed information about the oral phase of eating, and the fine co-ordination of breathing and swallowing, which has certainly contributed to our understanding of the oral and pharyngeal difficulties of children with cerebral palsy. Objective measures of these processes are to be welcomed.

We feel that the paper also raises several questions concerning the possible role of EDAT in clinical assessment. After reporting EDAT data, the authors go on to propose factors which may contribute to the observed difficulties, suggesting, for example, that the abnormal respiratory patterns observed in the anticipatory phase are due to impaired sensory perception. However, there is no clear evidence or argument to support this. This type of speculation provides a questionable foundation for the often rather vague management suggestions e.g. 'involve other feedback systems', 'stimulate hunger' etc. No data are presented on the effects of the proposed management strategies. Unfortunately the effect of this speculation is to bring the credibility of EDAT into question.

In their conclusion the authors suggest that EDAT provides sufficient information to assess function in the phases of swallowing. Many descriptions of the children's skills reported in the paper, come not from EDAT but from clinical

observation (e.g. poor head posture). It is also suggested in the paper that EDAT could limit the use of videofluroscopy but no conclusions are presented on the relationship between the respiratory patterns observed and the risk of aspiration. Whilst we would accept the limitations of videofluroscopy, and would only recommend it when a clear history of chest pathology emerges from the medical history, we remain unconvinced that EDAT offers a useful alternative.

We look forward to further studies which explore the effects of management strategies proposed in this paper, and which further delineate the role of EDAT within the spectrum of assessment techniques available to clinicians working with this client group.

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'Selley and Tripp reply'

SIR–Cerebral respiratory centres are continuously receiving sensory feedback stimuli to regulate breathing and we believe they also contribute to the control of the very complex feeding respiratory pattern, immediately before and during swallowing a spoonful of drink. This hypothesis has been substantiated and reported previously.[1]

EDAT is a diagnostic technique using non-invasive equipment as an aid to clinical assessment. We note the writers' point regarding the anticipatory phase, which has received too little attention and is not assessed objectively by videofluoroscopy. We believe under the test conditions of feeding the participants (i.e. without verbal prompts) that sensory stimuli provided by vision and the interpretation of those sensations are the most likely afferent inputs to account for changed respiratory patterns in preparation for an inspiration during the delivery phase. The correspondents do not suggest an alternative, but other common possibilities are verbal instructions and proprioceptive feedback during assisted or self-feeding, which may be employed during management of feeding difficulties to help compensate for impaired visual perception i.e. 'involving other feedback systems'.

No conclusion was reached relating abnormal post deglutition inspirations to potential lung pathology because we could not find any evidence to substantiate that hypothetical risk in our series of 179 severely dysphagic children with cerebral palsy.

We would not hesitate to use videofluoroscopy in the rare situation your correspondents described. We have never suggested that EDAT could offer an alternative to a videofluoroscopic evaluation when clinically justified. However, the speed and complexity of the swallowing mechanism is so complex that an objective aid to a clinical assessment is valuable to help formulate an accurate diagnosis before advising on management.

We, too, look forward to further studies and are gratified by reading favourable reports from other centres in the UK and USA. The authors would be pleased to help if requested.

WG Selley
JH Tripp

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'HHH syndrome associated with callosal agenesis and disordered neuronal migration'

SIR–Whilst the association of some congenital errors of metabolism with CNS malformation is well recognised (e.g. polymicrogyria and pachygyria in Zellweger's syndrome and callosal agenesis in non-ketotic hyperglycinaemia), no such association has as yet been identified in hyperornithinaemia-hyperammonaemia-homocitrullinuria (HHH) syndrome. We report here on a child with HHH syndrome who also had callosal agenesis and a neuronal migration defect.

A 10-month-old girl born to consanguineous parents was referred because antenatal and neonatal ultrasonography had suggested callosal agenesis. Pregnancy had been otherwise uncomplicated and delivery was by elective Caesarean section at 37 weeks' gestation. The neonatal period was complicated by poor feeding and respiratory symptoms, which led to the identification of a sequestered segment of the right lower lobe. Following discharge from the neonatal unit she remained in good health and made normal developmental progress. She was described by her parents as a fussy eater who vomited frequently. Weight gain followed the 10th centile as did head growth. There was mild hypertelorism and mild hypotonia with no other abnormality on neurological examination.

MRI confirmed callosal agenesis and, in addition, identified extensive sub-ependymal nodular heterotopias and an area of left temporal polymicrogyria. Biochemical analysis revealed hyperammonaemia and hyperornithinaemia with borderline high levels of glutamine and alanine. Urine analysis detected homocitrullinuria and marked orotic aciduria. Fibroblast studies carried out according to the method of Gray and colleagues[1] *showed low ornithine uptake consistent with HHH syndrome.*

A low protein diet has produced both clinical and biochemical improvement. Genetic analysis is in progress to determine whether this patient has inherited any of the mutant ORNT 1 genes described by Camacho and colleagues.[2]

Over forty patients with HHH syndrome have been

reported.[3] The phenotype is variable. Symptoms are usually those of episodic hyperammonaemia which may pass undetected until adulthood. Hypotonia in infancy is described often progressing to spasticity in childhood. Cognitive abilities range from normal intelligence to severe learning difficulty. Only one patient has been reported as having an abnormality on neuroimaging. This adult described by Lemay and colleagues[4] had multiple foci of increased signal in his subcortical white matter which were interpreted as representing demyelination and were associated with a sensorimotor peripheral neuropathy. No patient, to our knowledge, has had an associated pulmonary anomaly.

The combination of a metabolic disorder and a structural brain abnormality in this child, along with the pulmonary anomaly, could represent the coincidental inheritance of two or more genetic disorders from consanguineous parents. However it may well represent another example of an association between a congenital error of metabolism and a congenital brain malformation and this re-emphasises the importance of metabolic investigation in the evaluation of unexplained cerebral dysgenesis.

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Book Reviews

Birth Counts: Statistics of Pregnancy and Childbirth. Volumes I and II
Edited by Alison MacFarlane and Miranda Mugford
London, The Stationary Office. 2000, Vol. 1 pp 447, Vol. 2 pp 673, Vol. 1 £29.95; Vol. 2 £29.95.
ISBN Vol. 1: 0 11 621049 4. Vol. 2: 0 11 620917 8

Birth Counts is a 'must have' reference for all those interested in child and maternal health. The two volumes bring together comprehensive statistics about pregnancy, childbirth, and new-born babies, and the socioeconomic circumstances in which they are born. The data reported are from those collected by government departments and the National Health Service in England, Wales, Scotland, and Northern Ireland. The first edition was written in the early 1980s and published in 1984. This second edition includes many but not all data published then, mainly concentrating on data for the 1990s with a shift of emphasis from cross-sectional data to time series.

Information relating to childbirth and infancy are the most frequently used health statistics and are essential for planning and monitoring both health and social services. Some of this information is available but is only found scattered in a variety of official publications or in some cases is never published at all. The value of having such data easily accessible in one source is immediately evident when reading this book.

The authors of this readable and informative book are Alison Macfarlane (National Perinatal Epidemiology Unit) and Miranda Mugford (University of East Anglia), working in close collaboration with statistical staff in the government statistical services of the UK.

The first volume provides a guide to the data contained in the second volume, describing how the data are produced in order to help the reader interpret them. By incorporating the historical background and developments over the years, the potentially dry facts are brought to life. This volume also provides help to deal with the possible factors which may influence the outcome of pregnancy i.e. the characteristics and circumstances of mothers and their babies and the care they receive. The care available and how this has varied over time is likely to be of interest to current service providers.

In chapters 8 and 9 the problems of definition and collecting suitable data about impairment, disability, and illness in childhood are discussed. The focus is mainly on the first year of life and morbidity in relation to birth and is less comprehensive, perhaps reasonably so, than previous chapters. Maternal mortality and reproductive health are covered in the next chapter. The costs of providing services, a sometimes neglected area, are of considerable interest to both families and the government. These are reviewed in chapter 11. Finally, there is a brief overview of comparisons between the outcome of pregnancy and childbirth in different countries.

The appendices to Volume I are as comprehensive and detailed as the main body of the book providing contact names and addresses or telephone numbers of data sources as well as official reports and classifications and – maintaining

the historical and legal perspective of the book – a chronological table of events and legislation.

Volume II is slightly larger and contains all the compiled data referred to in Volume I, in table form.

Anyone working in maternal and child health will find something of interest on almost any page in this book. I would recommend it for the library of every maternity, neonatal, or child health unit. Many practitioners will also want a copy ready-to-hand in their office.

Alison Salt

The Chailey Approach to Postural Management
By Teresa E Pountney, Catharine M Mulcahy, Sandy M Clarke and Elizabeth M Green
Birmingham, Active Design. 2000, pp 157, £25.00
ISBN 0-9538262-0-1

This book describes an approach used at Chailey Heritage Clinical Services in East Sussex, UK, to provide assessment and treatment of children requiring postural control intervention due to neurological impairment. The authors use the principles of developmental biomechanics to address the postural abilities of the children and to prescribe postural management equipment as a link to the child's fine motor and cognitive skills. Using a training manual format, self-teaching sections are provided that create a user friendly text with quality illustrations, review questions, and assessment charts to assist in the learning process. The content is developed for an audience with many levels of expertise and experience by incorporating basic information about postural abilities as well as more advanced theories of biomechanics and motor control.

The first chapter introduces the reader to definitions and the nine components used to analyze the Chailey levels of postural abilities in children. This foundation provides the basis for analysis of the child's posture and movement abilities in 5 positions presented in Chapter 2: supine, prone, sitting (floor), sitting (box), and standing. Illustrations are provided to identify different levels of ability in the different positions. These principles of children's postural control are then related to functional skills in the next chapters.

The fourth chapter includes a review of the knowledge base in biomechanics (forces) and muscle structure (fibers, length) for problem solving musculoskeletal changes and bone adaptations observed in children with neurological differences. In addition, the authors incorporate concepts of neuroplasticity, sensory processing, and motor control and motor learning as links to increasing motor and cognitive skills of the child.

The remainder of the book addresses the assessment process and planning postural management for the child. The authors should be commended for their individualized approach to addressing the postural abilities of children with emphasis on 24 hour management and experiential learning of the client. They outline the importance of clinical reasoning to address individual postural differences through three 'tracks'. The Procedural Track is used to analyze the child's developmental level of postural control, the Interactive Track identifies the postural needs of the specific child to effectively interact with key persons and objects in the environment, while the Conditional Track of clinical reasoning addresses issues specific to the values of family, the potential outcomes of the child, and analyzes the resources available to support the goals of the family, child, and therapist.

The systematic approach the Chailey group provides in this book will be helpful to students as well as practicing clinicians who problem solve postural management of children with neurological and other impairments. The illustrations, assessment charts, and clinical reasoning approach make this publication a unique comprehensive resource in this important area of intervention for children.

Patricia A Burtner

Early Diagnosis and Interventional Therapy in Cerebral Palsy: An Interdisciplinary Age-Focused Approach (3rd edn)
Edited by Alfred L Scherzer
New York, Marcel Dekker, 2000. pp 355, US$55.00
ISBN 0 8247 6006 9

Professor Scherzer's latest edition of *Early Diagnosis and Interventional Therapy in Cerebral Palsy* is to be welcomed. It gives a good overview of the early years of a child with cerebral palsy together with earlier chapters on definition, classification and important trends in the changing epidemiology of the condition. There are informative reviews of habilitative services and a step-by-step approach to the assessment and complex treatment planning required for any child with cerebral palsy. These will be of considerable value to a child development team who will benefit from a template upon which to design or modify work in this area.

The third edition of this book acknowledges the enormous advances (although there is still a long way to go) in the understanding of the basis of cerebral palsy and in the management of a child with this condition. Having mentioned the issues of, for example, botulinum toxin, intrathecal baclofen, and dorsal rhizotomy in the preface, I was a little disappointed to find only minimal mention of these issues in the main text. Although the book is written to focus on the 0 to 3 year age group, these techniques are likely to rise (or fall) over time in their level of appropriateness for children of this age.

The review of research and treatment outcomes by Charlene Butler are detailed and informative, but readers should not go looking for the answers about outcomes because they are not there. More often than not we can say only that lack of evidence cannot be construed as negative evidence and we continue to be dogged by methodological difficulties in evaluating intervention in cerebral palsy. Nonetheless anyone considering embarking on a study designed for intervention would do well to consider these chapters carefully. My only significant criticism of this text is the scant reference to cerebral palsy in the developing world; this issue is of such importance, if only in terms of sheer numbers, that surely it warrants its own chapter.

Martin Kirkpatrick