(eg, admitting the child and taking a scan in the morning if symptoms have persisted for a threshold period after injury) improve the positive predictive value sufficiently to overcome economic and radiation-exposure concerns?

The authors correctly state that their study has established a rule that now needs prospective validation in further studies. Previous studies of the implementation of the Royal College of Surgeons head injury guidelines found

that they were not being fully followed: recommended numbers of SXR, computed tomograms and admissions were not occurring because clinicians believed them to be inappropriately aggressive. A robust validation of CHALICE discussing these more complex cost- and risk-benefit analyses will be required to reassure clinicians and ensure that history does not repeat itself.

Arch Dis Child 2006;**91**:877-878.
doi: 10.1136/adc.2006.104513

Correspondence to: R Forsyth, Department of Child Health, The Royal Victoria Infirmary, Queen Victoria Road, Newcastle upon Tyne, NE1 4LP, UK; r.j.forsyth@newcastle.ac.uk

Competing interests: None declared.

REFERENCE

- 1 Dunning J, Patrick Daly J, Lomas J-P, *et al*. Derivation of the Children's Head Injury Algorithm for the prediction of Important Clinical Events decision rule for head injury in children. *Arch Dis Child* 2006;**91**:885-91.

Training

Paediatricians subspecialising in cardiology: clinical governance, maintenance of expertise, and training

J L Gibbs

Perspective on the paper by Pushparajah *et al* (see page 892)

The inability of the limited workforce of paediatric cardiologists to cope with an ever increasing workload and the need for more paediatricians with some expertise in cardiology to address this problem has been highlighted in this issue by Pushparajah and colleagues.¹ Paediatricians undertaking such a role, whether in district general hospitals or teaching hospitals are likely to be undertaking most of their cardiology practice on their own, potentially placing themselves in a vulnerable position. There is a need to establish clearly the potential roles of these posts for the protection of both doctors and patients. Level of expertise will inevitably vary according to the training and aspirations of individual consultants, but this should not prevent all those involved adhering to certain common standards. The British Congenital Cardiac Association (BCCA) and the Royal College of Paediatrics and Child Health (RCPCH) have recently endorsed a brief guide (see *ADC* website: www.archdischild.com/supplemental) to practice for paediatricians with a special interest in cardiology, initially introduced for governance purposes in the Yorkshire, Humberside and Northern Trent paediatric cardiac network after local consultation with doctors and patients. The guide was heavily influenced by the Department of Health Paediatric and Congenital Cardiac Services Review

(PCCSR, published in response to the Bristol Inquiry Report).² Other than generic recommendations for local audit and joint practice with cardiologists from a tertiary centre, the guide recommends that any child deemed to have a cardiac abnormality should see a cardiologist on at least one occasion, and that each child should have a clear plan for follow up, jointly agreed by cardiologist and paediatrician. While paediatricians more experienced in cardiology may view such a recommendation as unnecessarily restrictive (particularly for simple anomalies such as small muscular ventricular septal defects), it is hard to argue against the PCCSR's recommendation (guided by the Children's Heart Federation) that any child with a cardiac abnormality should have access to a specialist opinion. This simple approach has the added value of minimising potential for iatrogenic cardiac disease.

In Pushparajah's practice every echocardiogram performed is reviewed at a later date by a cardiologist, which appears to work well for the individuals concerned. The BCCA/RCPCH guide does not see that as essential, but does make firm recommendations for documented audit on an ongoing basis to ensure that quality is maintained, and that appropriate measures for CPD and maintenance of expertise (usually by regular periods of secondment to a

tertiary cardiac centre) are in place. These measures demand a formal approach, agreed within job plans and with managerial and financial support from NHS Trusts.

It is important to recognise the major role played by cardiac technical staff in supporting "outreach" cardiology. The BCCA, in liaison with the British Society for Echocardiography (BSE) are currently working together to develop guidelines for practice for technical staff, including a mechanism for formal BSE accreditation in paediatric echocardiography for those working outside tertiary congenital heart disease centres. These guidelines, to be piloted later this year in several UK centres, will be based on the current BCCA/RCPCH recommendations for paediatricians practising cardiology but will include modern methods of assessment of competence such as direct observation of practical skills (DOPS), recently introduced for physicians in higher specialist training.

Training opportunities for paediatricians wishing to subspecialise in cardiology as consultants are, happily, expanding. NHS Trusts and their funding bodies are beginning to appreciate the benefits of such skills and there are now specifically designated training posts in most UK cities. These posts are open to trainees already in established general paediatric training posts, are advertised nationally, and are appointed in open competition. The posts are for one year, are administered by the tertiary cardiac centre's Postgraduate Deanery, and are specifically funded, the trainee returning to their original training scheme at the end of the year. If current progress with funding training can be matched by funding of consultant posts for paediatricians with cardiology as their subspecialty, quality of local care for children with suspected or confirmed heart disease will be hugely improved.

Arch Dis Child 2006;**91**:878-879.
doi: 10.1136/adc.2006.099283

Additional material is available on the ADC website (<http://www.archdischild.com/supplemental>)

Correspondence to: Dr J L Gibbs, East Floor Jubilee Wing, Leeds General Infirmary, Calverley Street, Leeds LS1 3EX, UK; jgibbs@boltonblue.com

Competing interests: None declared.

This document was unanimously supported at the Yorkshire, Humberside and Northern Trent paediatric cardiac network meeting held in York in March 2005. It was supported and adopted on a National basis by the British Congenital Cardiac Association Council in May 2005 and by the Royal College of Paediatrics and Child Health Executive Committee in April 2006.

REFERENCES

- 1 Pushparajah K, Garvie D, Hickey A, *et al*. Managed Care Network for the assessment of cardiac problems in children in a district general hospital: a working model. *Arch Dis Child* 2006;91:892–5.
- 2 Paediatric and Congenital Cardiac Services Review report. Department of Health, December 2003. www.advisorybodies.doh.gov.uk/childcardiac/reports.htm.

Frequency of metabolic disorders

Frequency of metabolic disorders: more than one needle in the haystack

P J Lee, P Cook

Perspective on the paper by Sanderson *et al* (see page 896)

Since Archibald Garrod coined the term “inborn freaks of metabolism” after his classic studies on alkaptonuria, cystinuria and pentosuria a century ago,¹ clinicians have believed that these disorders are a rarity. Early progress was hampered by insensitive methods of detection, as well as a lack of awareness within the medical community as a whole. The successive application of chromatographic techniques, mass spectrometry, enzymology and, recently, molecular genetics has increased our ability to detect metabolic perturbations in symptomatic, and in asymptomatic, people. Despite possible diagnosis, nihilism persisted as few therapeutic interventions were available and many of those affected died early in childhood. Even with survival, quality of life was extremely poor.

Phenylketonuria (PKU) is a good example of how the application of new advances, diagnostically and therapeutically, can have a dramatic effect on outcome and therefore on expectations. PKU was first described by Følling in the 1930s. Biochemical diagnosis could be made on urine samples from infants with developmental and neurological problems. Two decades passed before Bickel showed that a phenylalanine-restricted diet could be given safely to affected children, although they remained severely impaired and dependent, with many of them institutionalised for life.² It required the development, by Robert Guthrie in the 1960s, of a cheap, sensitive and specific screening test that could be applied to large populations before a true effect on outcome could be shown.³ Since then,

further technological improvements have provided accurate, quantitative measurements of phenylalanine, and an explosion of phenylalanine-free nutritional products means that a newborn with PKU diagnosed today can expect a near-normal life. Management of PKU is certainly one of the success stories of 20th century medicine.

PKU is well known to paediatricians, but many other inherited metabolic diseases (IMDs) are not. *The metabolic and molecular bases of inherited disease* edited by Scriver *et al*⁴ lists so many IMDs that it has necessitated an existence on the internet rather than on paper. The use of laboratory techniques such as tandem mass spectrometry to measure acyl carnitines, gas chromatography–mass spectrometry to measure organic acids, tissue enzymatic tests to measure lysosomal, intermediary metabolic and respiratory chain enzymology, and molecular genetic testing mean that many of these disorders can be readily detected. Once a diagnosis is made, other family members at risk can be screened; appropriate genetic counselling about future pregnancies can be offered; and treatment, where available, can be introduced. The main question remains as to whether or not clinicians are sufficiently aware of the possible presentations to consider the diagnosis of an IMD. As these conditions have been considered to be rare, many clinicians will not have come across IMDs during their training or practice. In this issue, Sanderson *et al*⁵, in a careful retrospective review of one region in the UK, have shown that the frequency of these conditions may be

more common than was previously estimated: 1 in 800 live births rather than the often quoted 1 in 2500–5000.⁵ Although the population they studied has a high proportion of ethnic minority groups in which consanguinity is commonplace, their reliance on just a single regional specialist laboratory may mean that their prevalence figures are an underestimate. It is therefore reasonable to extrapolate their figures, suggesting roughly 800 new cases of IMD diagnosed each year in the UK.

This figure is not insignificant, as treatment for many of these conditions has improved considerably, increasing the survival rates and quality of life. Examples include enzyme replacement therapy for Gaucher's disease and other lysosomal storage diseases, with 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione for tyrosinaemia type I, sodium benzoate and sodium phenylbutyrate nitrogen scavengers in urea cycle defects; overnight glucose pump feeds and uncooked cornstarch in hepatic glycogenoses; and organ transplantation such as bone marrow for leucodystrophies or liver transplantation for amino acidopathies. Preliminary data from the newborn screening trial for medium-chain acyl CoA dehydrogenase deficiency in the UK suggest that mortality and morbidity can virtually be eliminated. Without screening, 20% of patients are expected to die at presentation, and a further fifth may survive but with considerable neurological damage.⁶ Children diagnosed with IMDs are surviving, but more adults are now diagnosed with these disorders for the first time. The data of Sanderson *et al*'s⁵ suggest that more than a quarter of patients are diagnosed at >14 years of age, sometimes even as late as in the eighth decade. Doctors need to be aware of these disorders.

All of this has considerable implications for services to provide care for these patients and their families. A recent UK survey by the same group identified at least 10 000 children and adults with IMDs in contact with current specialist services in the UK.⁷ Using the old prevalence figures, the group estimated that at least 8000 other