school students find work experience difficult to obtain. Some recall having to contact as many as 50 doctors, often with no response. Exposure to the workings of a hospital not only allows an appropriate student to obtain the credentials for shortlisting for interview but will also inform some that this is not the career path they had envisaged.

In our experience, 3 days provides a reasonable opportunity for observation and the chance to talk to a variety of allied health providers including nurses, technicians, physician associates, junior doctors and medical students.

The Royal College of Physicians (RCP) have recently indicated a wish recognise the contribution of consultants to the education of current medical students. We would argue that facilitating the selection of appropriate students to enter medicine is equally important.

There is no obligation for consultants to perform this role, the rewards being philanthropic. Recognition by the RCP would encourage other consultants to participate and would help inform the next generation of potential medical students. ■

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Response

The RCP very much recognises the value of the medical educator, often not reflected in a job plan. Consultants do undertake this activity as part of our professional role at undergraduate and postgraduate levels and provide educational and work experience for school children and other students. In my experience it is common practice for trusts to have widening participation officers / work experience coordinators to facilitate ease of access for aspiring medics. This adheres to the widening participation agenda. Consultants generally are cooperative and happy to help and don't expect a reward, as is the case with other members of staff. However, we are exploring how more formal recognition of the educator role may be augmented. ■

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Not only what investigations? When, why, at what cost, whose choice?

Editor - Regarding the recent publication in *Clinical Medicine* by Vasudevan and Suri,[1] I was pleased to see the subject but very disappointed in this article. As a member of the Royal College of Physicians and Royal College of Paediatrics and Child Health, I felt very saddened that neither a paediatrician nor a service user contributed to this article. There is imprecision in the definition of terms, leaving out important criteria such as how delayed is 'significant' in global developmental delay (GDD); it is usually defined as >2 standard deviations below the mean.[2–4] In addition, GDD is referred to as if it is just intellectual disability (ID) in young children – except where DD is severe, the development of future intellectual disability is uncertain.

The World Health Organization's definition of mental retardation and the Department of Health (DH) definition of learning disability are clearer: they include mention of IQ (<70) and degrees of disability that also relate to the likelihood of finding a cause. The DH definition of learning disability is misquoted (by partial omission) and the reference to it leaves out the important distinction therein between general and specific learning disabilities (eg dyslexia).[6] There is little reference to the evidence base informing practice or to patients' views.

No mention is made of the ethical and practical considerations of testing and of obtaining valid consent, nor of the considerable variation in guidance and practice in investigations for GDD and ID. These issues are mentioned for future developments such as whole-genome sequencing but are very relevant for current investigations.

Considerable distress can be caused by blood tests, waiting for results and the frequent occurrence of copy number variations of uncertain significance.[8] In my experience, parents' wishes vary: many want to investigate their child's disability exhaustively and immediately, most do not, and some want no investigations unless likely to have a significant impact on treatment such as thyroid function.

Microarray has not been routinely available for 2 decades – at least not from the Guy's and St Thomas' genetic department in London and I guess not many other places either. The recommendation of magnetic resonance spectroscopy as routine seems unjustified – there is evidence of little additional diagnostic yield. If properly audited, part of research projects would be more appropriate.

I do recognise that better investigations do identify a cause more commonly, and rarely but increasingly identify treatable conditions. Nonetheless, even a specific diagnosis most often does not help the individual very much if at all. The comment that a specific diagnosis enables access to special education and social care is sometimes true but is against the spirit and text of the relevant law, which for children at least has been based on identification of needs not diagnosis since the Education Act 1981, and the Children Act 1989.

Participation is key in working long term with patients and their families with significant learning difficulties. For children, parents usually give consent for their child. They may have very different priorities and concerns to the clinician. Respectful explanation of options, uncertainties, costs and benefits of investigations is vital for valid consent. This remains an art[7] and does not fit well with a mandatory list of investigations all at once for all.

Adults with ID may not be competent to give valid consent. Surely this should at least be mentioned, and references given to how to address this, both respecting the autonomy of the individual and not neglecting their health needs.

There are, coincidentally, far better recent articles, and discussions of the pros and cons of aetiological investigations for GDD and ID – see references 2–5 below, which of course do not totally agree with one another. ■

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References

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Response

Thank you for your thoughts on our review of an approach to patients with global developmental delay (GDD) and intellectual disability (ID). We were invited to write this article, pitched for physicians (not specialist paediatricians or clinical geneticists) seeing young and older adults with GDD/ID. A generic diagnostic approach to understanding the myriad of causes of GDD/ID is difficult to find due to the non-discrete spectrum between mild, moderate and severe, across multiple domains and this is often a challenging area for general physicians. Our review aimed to help a physician think about ID/GDD in two contexts: firstly, could an identified inherited susceptibility to developmental delay provide insight into future wider health needs and secondly, how the approach to genetic testing in this context is changing. Addressing the points raised in this letter to the editor, the DH definition of learning disability is not misquoted by us. The reference was provided so that readers could read the entire report for more detail. We concur that ethical issues around consent for testing are important. While some families may not seek a diagnosis for their child's developmental delay, our experience suggests that the majority do. Our experience also suggests that families who have children with unexplained GDD/ID often return to clinical genetics services for a diagnosis when their affected children are adults. This may help to inform risk of recurrence of similar problems in children of unaffected siblings and sometimes for parents to obtain closure. There is no doubt that improved diagnostic and (through the medical literature) prognostic information is often extremely helpful to patients and families in accessing services and healthcare. While all children with GDD should be able to access special education and social care, in practice limited resources means that children with GDD with a specific underlying diagnosis are more likely to be able to access these services than children with GDD without one. In addition, a molecular diagnosis is often key to accessing research studies and to predict the development of any future medical complications. References 4, 5, and 7 here discuss the evidence-based recommendations on investigating children with GDD. These articles were all published in the same issue of Archives of Disease in Childhood in November 2017, whereas our review was submitted for publication in May 2017. Chromosomal microarray has been available to patients in Nottingham initially through a research project from the Wellcome Trust Sanger Institute (0.5 Mb BAC array) from 2006 and on a service basis from the Regional Cytogenetics Laboratory from 2009. We have recommended that MRI brain and MRS should be undertaken in patients with GDD/ID who have microcephaly or macrocephaly or abnormal neurological findings, not as a routine investigation. To conclude, we have attempted to summarise an approach to GDD/ID, highlighting that next-generation sequencing in the form of trio whole exome sequencing or whole genome sequencing will likely result in an increasing diagnostic yield in patients with GDD/ID. ■

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Editorial note

We would like to apologise to the authors of *Diagnosis and management of cerebral venous thrombosis* (*Clin Med* 2018;18:75–9.) for incorrectly listing their job titles and affiliations in the printed version of *Clinical Medicine*. The correct listing is present on all online records of this article.