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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.