



AB008. Precision medicine in neurodevelopmental disorders: optimising outcomes for children and families through timely, accurate diagnosis and targeted treatment

Simone Ardern-Holmes^{1,2}

¹TY Nelson Department of Neurology and Neurosurgery, The Children's Hospital at Westmead, Westmead, NSW, Australia; ²Faculty of Medicine and Health Sciences, The University of Sydney, Sydney, NSW, Australia

Correspondence to: Simone Ardern-Holmes. TY Nelson Department of Neurology and Neurosurgery, The Children's Hospital at Westmead, Westmead, NSW, Australia; Faculty of Medicine and Health Sciences, The University of Sydney, Sydney, NSW, Australia.

Email: simone.ardernholmes@health.nsw.gov.au.

Abstract: Neurodevelopmental disorders including developmental delay, intellectual developmental disorders, and autism, with comorbidities such as epilepsy, occur

frequently in children (3–5 percent). Careful phenotyping and individualized investigations are needed to ensure accurate diagnosis and early identification of acquired or genetic causes. Specific management is indicated in many cases, to optimise patient outcomes and prevent recurrence. An approach to clinical assessment will be outlined, highlighting important features on examination, and neuroimaging. Resources to inform the diagnostic process will be presented, emphasizing the early identification of treatable causes of neurodevelopmental disorders, and suggested criteria for referral to sub-specialist neurologist, metabolic physician and clinical geneticist. A comprehensive approach to management of the child and family is outlined, to improve outcomes and quality of life. Instructive case examples will be provided of single gene disorders for which specific targeted treatments are available, illustrating the potential to change the natural history of these conditions.

Keywords: Neurodevelopmental disorders; diagnosis; targeted treatment

doi: 10.21037/pm.2020.AB008

Cite this abstract as: Ardern-Holmes S. Precision medicine in neurodevelopmental disorders: optimising outcomes for children and families through timely, accurate diagnosis and targeted treatment. *Pediatr Med* 2020;3:AB008.