

Developmental regression: Always a worrying signal

Daniel Keene MD FRCPC¹, Danielle Grenier MD FRCPC²

An 11-month old girl is presented to you for developmental delay. The child started to roll over at four months of age and sit independently at approximately seven months of age. Afterwards, she gradually regressed and could no longer achieve these motor milestones. In the past few weeks, she lost the ability to hold her head straight or bear weight on her legs. Her babbling and smiling have decreased, and she does not seem to be interested in her surroundings. The pregnancy and delivery were uneventful, and there is no history of neonatal complications, febrile illnesses or head injuries. The family reports that she received her first three pentavalent immunizations. An analysis of family history was negative for developmental delay; however, the parents are distant cousins.

On examination, the child was unable to sit, roll or bear weight on her legs. Her eyes fixed poorly on objects and she did not reach for them. Her trunk and limb muscular tone was poor, while the muscle bulk remained normal. Deep tendon reflexes were hard to obtain. In view of the developmental deterioration and abnormal neurological examination, the patient was referred to a paediatric neurologist.

LEARNING POINTS

- Primary health care providers treating children with progressive intellectual and neurological deterioration (PIND) are facing a challenging task, because a myriad of neurodegenerative disorders can present in a similar fashion.
- Regression of developmental milestones or PIND should alert primary health care providers of the need to conduct a detailed history and physical examination.
 - When clinically indicated, investigations such as magnetic resonance imaging, electroencephalogram, evoked potentials, biochemical studies, genetic investigations (cytogenetic studies and molecular testing) and biochemical evaluations should be considered.
 - A definitive diagnosis may require further investigations and more invasive testing (1,2).
- The two-year surveillance study (3) on PIND was used to enhance monitoring for the possibility of Creutzfeldt-Jakob disease (CJD) and variant-CJD. While CJD is rare, the occurrence of variant-CJD in Canada would signal an important change in its epidemiology.
- Canadian Paediatric Surveillance Program participants reported 99 possible PIND cases; 59 of these were confirmed. The three most frequent diagnoses were mitochondrial disorders, ceroid lipofuscinosis and mucopolysaccharidosis. One case of iatrogenic CJD was confirmed; however, no cases of the variant form were reported. Of note, there were no cases with toxic etiologies (4,5).
- A definitive etiological diagnosis could not be attained in eight cases, even after exhaustive investigations and review by an expert panel consisting of four paediatric neurologists, a medical geneticist and a paediatric neuropathologist.
- Support for ongoing research and surveillance on PIND disorders is essential to advance knowledge and provide useful public health information about trends occurring in rare neurological disorders.

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The Canadian Paediatric Surveillance Program (CPSP) is a joint project of the Canadian Paediatric Society and the Public Health Agency of Canada, which undertakes the surveillance of rare diseases and conditions in children and youth. For more information, visit our Web site at <www.cps.ca/cpsp>.

¹Department of Paediatrics, Division of Neurology, Children's Hospital of Eastern Ontario; ²Canadian Paediatric Society, Ottawa, Ontario
Correspondence: Canadian Paediatric Surveillance Program, 2305 St Laurent Boulevard, Ottawa, Ontario K1G 4J8. Telephone 613-526-9397 ext 239, fax 613-526-3332, e-mail cpsp@cps.ca
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