Hypotonia in a newborn: Unravelling more than expected

A 30-year-old healthy primigravida delivers a daughter at 35 weeks' gestation. Her pregnancy was uneventful except for decreased fetal movements. At birth, the infant was hypotonic and grayish, with poor respiratory efforts, and had clubfeet. Apgar scores were 4 and 6 at 1 min and 5 min, respectively. The infant required intubation and mechanical ventilation for a total of 32 days. Postextubation, she had many apneic spells and feeding difficulties. Nasogastric reflux was confirmed, and she underwent fundoplication and gastrostomy. In view of the hypotonia, as well as the myopathic facial appearance of the mother, the neonatologist suspected congenital myotonic dystrophy (CMD). He ordered chromosomal studies, and confirmed that mother and daughter had myotonic dystrophy.

LEARNING POINTS

- Between March and December 2005, the CMD study of the Canadian Paediatric Surveillance Program has confirmed three cases.
- Myotonic dystrophy is an autosomal dominant, multisystem disorder caused by a genetic mutation – an unstable expanded CTG repeat sequence.
- The phenomenon of 'genetic anticipation' occurs in CMD, where a more severe clinical phenotype and earlier age of onset can occur over subsequent generations.
- Frequently, the infant with CMD is the 'index case', with the mother's diagnosis detected after the birth.
- Neonatal mortality rates range from 17% to 41%. Recent evidence indicates that infants can survive after prolonged ventilation (longer than 30 days), affecting management decisions.
- Approximately 66% of children with CMD have delayed cognitive development. Many may also have learning disabilities, attention deficit hyperactivity disorder and aspects of pervasive development disorder. Children benefit from early introduction of therapeutic playgroup, physiotherapy and ongoing multidisciplinary care.

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