

Fragile bones or child maltreatment?

A nine-month-old boy with a swollen leg is brought to the emergency room by worried parents. They report a fall earlier during the day while the infant was pulling to stand and irritability thereafter. The family history is negative for congenital bone fragility conditions. An in-depth review of the social situation does not suggest any risk factors for abuse. Physical examination reveals a well-nourished infant with slightly blue sclerae, mild anterior bowing of both legs and a swollen, tender right lower leg. A radiograph confirms a displaced, mid-shaft right tibial fracture but no evidence of rickets. A skeletal survey shows

multiple wormian bones in the cranial sutures, vertebral compression fractures and thin, gracile long bones. A bone mineral density study at the lumbar spine is 1.8 standard deviations below the mean. Even though no mutations in the type I collagen genes are identified, osteogenesis imperfecta (OI) type IV is diagnosed on the basis of clinical severity and radiological studies. The tibial fracture is treated with external reduction and casting. After one year of intravenous cyclical pamidronate, there are no further fractures, and reshaping of the vertebral bodies is evident radiologically.

LEARNING POINTS

- The OI study was added to the CPSP to raise awareness of novel OI forms and to document the geographical distribution. In the first year, 13 cases were confirmed in children ranging in age from 0.3 to 14 years (median 5.1 years). Eight of these cases were from Ontario.
- OI is usually caused by mutations in type I collagen genes (*COL1A1* and *COL1A2*), with at least seven types identified leading to fragile bones.
- At diagnosis, children presented with five fractures (n=4), five to 10 fractures (n=6), and 10 fractures (n=3).
- Classical OI diagnosis is straightforward but may be challenging in the absence of affected family members. Parents of children who have milder forms of OI and/or in whom OI presents 'de novo' with low-trauma fractures may be falsely accused of child abuse.
- Types of fractures typically observed in both child abuse and OI include rib fractures, spiral fractures, fractures in multiple stages of healing, and fractures for which there is no adequate explanation of trauma.
- A diagnosis of OI does not preclude the possibility of physical abuse, and both diagnoses should be considered. Comprehensive review of clinical status, plausibility of the reported mechanism for the injury and risk factors for abuse should be part of the initial evaluation.
- A consultation with a geneticist familiar with OI may reveal a family history of mild OI if symptoms such as blue-tinted sclerae, presenile hearing loss, rickets, dental problems, short stature and/or a history of broken bones are identified.
- Diagnosis of OI in infancy or early childhood allows a window of opportunity for starting multidisciplinary care and bisphosphonate treatment leading to disappearance of bone pain, improvement of mobility and diminution in the numbers of fractures.

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>.