

Child abuse and/or osteogenesis imperfecta? The challenging management of patients with unclear diagnosis

To the Editor: A young mother presented to the trauma unit at Red Cross War Memorial Children's Hospital, Cape Town, South Africa (SA) with her 7-month-old female baby, who had a large left-sided parietal swelling. She reported that the baby had fallen from her arms when she was exiting a minibus 4 days earlier. However, she claimed that she had only noted the swelling on the day she presented at our hospital. There was no history of loss of consciousness, seizures or vomiting. The baby was playful and feeding well.

On physical examination, a large and fluctuating left parietal scalp haematoma was detected. A computed tomography scan of the brain showed three parietal skull fractures: an acute fracture with significant overlying scalp swelling and underlying subdural haemorrhage, and two older fractures without scalp soft-tissue swelling (Fig. 1). The baby was admitted to the trauma ward for neurological observation and a multidisciplinary non-accidental injury (NAI) work-up. A skeletal survey revealed four more fractures: a bilateral healed tibial corner fracture (Fig. 2), an older right femoral metaphyseal fracture (Fig. 3), and a wedge compression fracture at the height of T11 (Fig. 4). Retinoscopy revealed no evidence of retinal haemorrhage. After admission we noticed that the baby had grey-blue sclera, in particular when compared with her mother (Fig. 5), which can be a spot diagnosis for osteogenesis imperfecta (OI), a rare genetic disorder associated with abnormal synthesis of type I collagen resulting in an increased risk of fractures. We consulted the genetics department at the hospital. They noted other typical symptoms of OI, including short stature and relative macrocephaly.

During the interview with the social worker, the patient's parents vehemently denied any sort of abuse or neglect. The Department of Social Development visited the patient's home and interviewed the people living on the premises, concluding that it was a suitable home

for the baby. We reported the case to the local police authority and an investigation was opened, but the prosecutor determined that there was no reasonable prospect of success and the case was closed. Under our care the baby was smiling and playful, showing no signs of developmental delay or lasting damage from the intracranial bleeding that she had sustained. After the Department of Social Development had deemed the family's care appropriate for the child and there was no further action from the police, the patient was discharged from the trauma ward. We followed her up closely, initially weekly and later monthly, to monitor for any new injuries. Clinical examination during these follow-up visits did not suggest any new injuries. Eventually she was lost to follow-up.

It can be difficult to distinguish between symptoms of OI and child abuse. The clinical picture of seven fractures, including two corner fractures and a subdural haemorrhage, in a non-walking infant aged 7 months, described above, indicated that abuse was the most likely origin of the injuries. NAIs are frequently seen in paediatric trauma

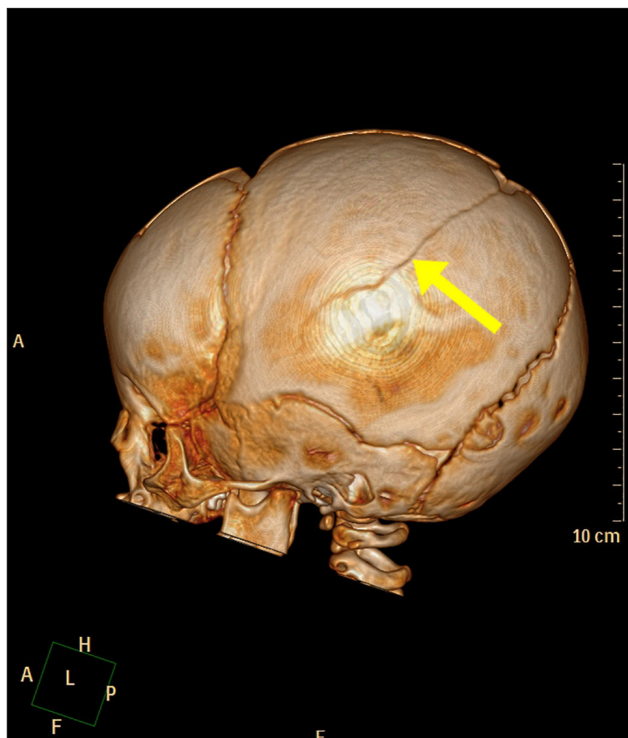


Fig. 1. 3D reconstruction image of a computed tomography scan of the head, demonstrating a linear parietal skull fracture on the left (arrow).

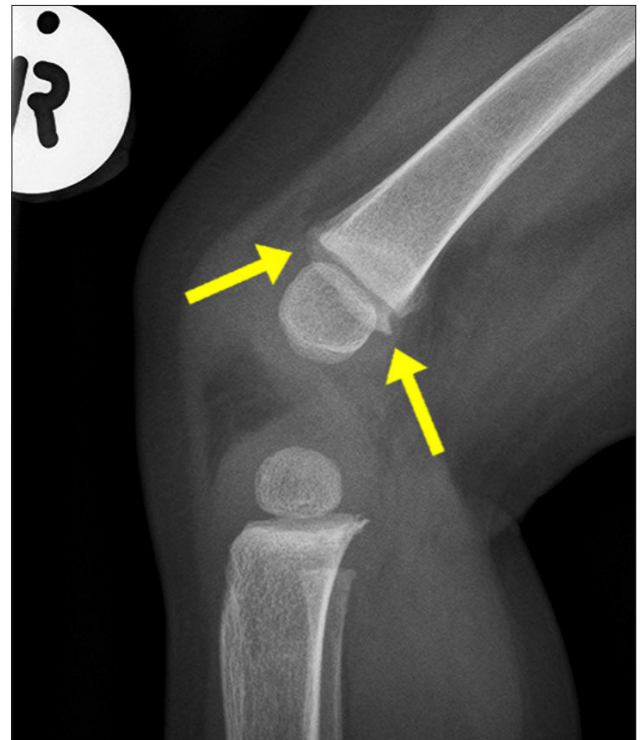


Fig. 2. Lateral radiograph of the right knee, demonstrating a corner fracture of the proximal tibia (arrows).

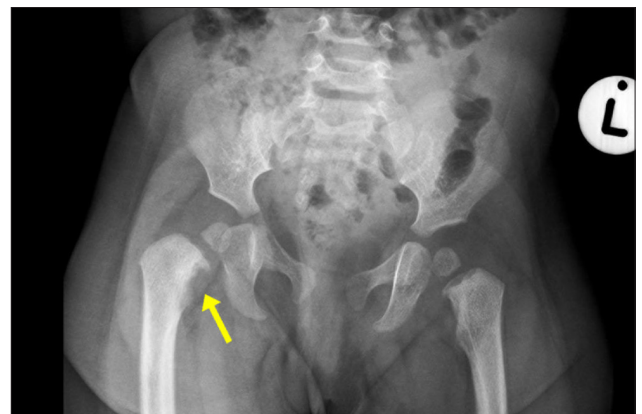


Fig. 3. Anteroposterior radiograph of the pelvis, demonstrating an old right femoral metaphyseal fracture (arrow).

patients across SA. Red Cross War Memorial Children's Hospital treats ~350 children aged <13 years for suspected physical and/or sexual NAI per year.^[1] The Optimus Study on Child Abuse, Violence and Neglect in South Africa reported lifetime rates of 34% for physical abuse among 15 - 17-year-olds.^[2] The World Health Organization (WHO) estimated that 95 million children across the globe experience abuse, with the highest rates being reported in the Africa region.^[3] The paucity of data, the shortage of resources, and lack of awareness regarding child abuse are major threats to achieving the United Nations Sustainable Development Goals (SDGs) on the African

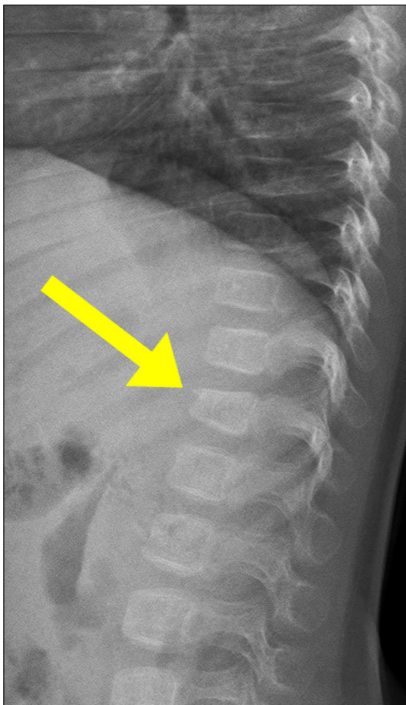


Fig. 4. Radiograph of the lateral vertebral column, demonstrating a wedge compression fracture at the height of 11th thoracic vertebra (arrow).

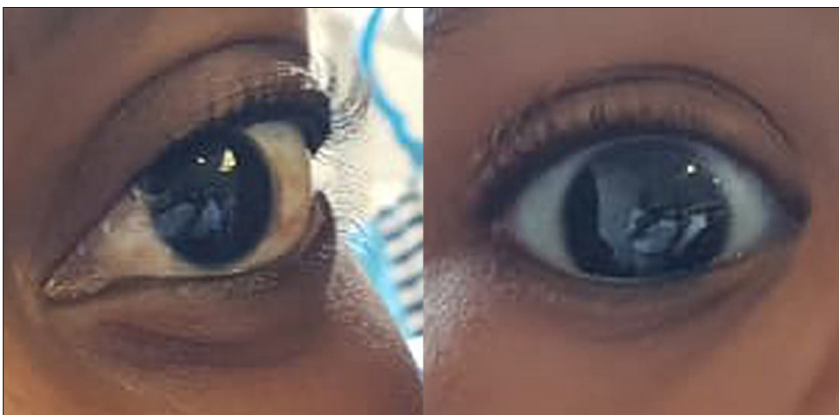


Fig. 5. Right eye sclera of the child (right) compared with the left eye sclera of her mother, showing the clear bluish discoloration.

continent. SDG 16.2 explicitly calls to end abuse, exploitation, trafficking and all forms of violence against and torture of children.^[4]

Clinical signs observed in our patient, including her short stature, relative macrocephaly and grey-blue sclera, were compatible with a diagnosis of OI. With OI, fractures of long bones are more common than corner or compression fractures. Bones often fracture from little or no obvious trauma. Subdural haemorrhage has been reported as a complication of OI,^[5] yet it is more often a result of NAI.^[1] OI should be considered as a differential diagnosis in children presenting with multiple fractures.^[6] At an early age, the symptoms of OI are not always obvious.^[6] At least eight different types of OI have been identified. Genetic testing for COL1A1 and COL1A2, the most common mutations associated with OI, can confirm the diagnosis, but is not routinely available for patients in SA. We were therefore unable to exclude the diagnosis with certainty.

OI and child abuse are not mutually exclusive. An underlying collagen deficiency, even if proven by genetic testing, does not in any way preclude neglect and/or abuse. This case illustrates how challenging the management of patients with a complex constellation of symptoms that allows multiple diagnosis can be. The case also emphasises the urgent need for more research on child abuse on the African continent in general, as well as the need for increased access to genetic testing, specifically for COL1A1 and COL1A2, to help doctors to distinguish between OI and NAI.

The management of child abuse cases is extremely difficult. If parents are suspected of abuse, how much access should they be allowed to the child? Importantly, clinicians have to keep in mind that removing a

child from a family and placing it into the hands of a foster care system that is already overburdened is traumatising for both child and parents, so these decisions require great scrutiny and a multidisciplinary work-up and discussion with the social workers and the investigating police officers. Doctors defending caretakers accused of abuse have testified in court, pleading – despite poor medical evidence – that a genetic condition had been overlooked.^[7] Irresponsible testimony is a danger to child wellbeing and a threat to our professional integrity as physicians. Clinicians should be cautious in their diagnosis, even when faced with enormous pressure.

In the case described here, despite intensive investigations and a multidisciplinary work-up including trauma, orthopaedics, radiology, ophthalmology, neurosurgery, genetics and social work, we were unable to come to a definitive diagnosis. Accepting that the full details of the patient's situation and the true facts may remain hidden forever was very difficult for the team.

First reports indicate that substantially fewer allegations of child maltreatment were reported during the first wave of the COVID-19 pandemic,^[8] while at the same time hospitals saw increased numbers of traumatic injuries caused by physical child abuse.^[9] The WHO has urged its member states to systematically address the suffering from child abuse and in other vulnerable groups during this period.^[10] Medical providers must be particularly attentive for signs of child abuse and/or neglect when evaluating patients at clinics and emergency departments in the context of the COVID-19 pandemic.

The ambiguity of this case highlights the importance of multidisciplinary teams of medical providers and social workers and emphasises the need for increased access to genetic testing for COL1A1 and COL1A2 in SA to help doctors to distinguish between OI and NAI.

Consent. Consent to publish was obtained from the patient's legal guardian and documented in the patient's medical records.

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