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Balancing Medical and Non-Accidental Causes of Multiple Fractures in a Child with Progressive Familial Intrahepatic Cholestasis

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
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Conflict of interest: None declared

Patient: Female, 2
Final Diagnosis: Hepatic rickets
Symptoms: Pain
Medication: —
Clinical Procedure: Skeletal survey
Specialty: Pediatrics and Neonatology

Objective: Rare disease

Background: All medical practitioners must be vigilant for child abuse and neglect (CAN) so that opportunities to intervene, prevent, and improve outcomes are not missed. However, child abuse is often overlooked in practice, and no sign or pattern of presentation of fractures is absolutely specific for child abuse.

Case Report: Here, we present the case of a 22-month-old girl with progressive familial intrahepatic cholestasis (PFIC) type 2 who presented with "red flag" fractures indicative of child abuse. Biochemistry showed vitamin D deficiency and a skeletal survey revealed rickets and multiple pathological fractures. However, her age, number of differently-aged fractures and their circumstances, and differential diagnosis of CAN prompted adherence to joint Royal College of Radiologists and Royal College of Paediatrics and Child Health guidelines for non-accidental injury.

Conclusions: This case highlights some important considerations in management of suspected CAN in the context of the pathophysiology of a rare hereditary disorder that can result in pathological fractures.

MeSH Keywords: Child Abuse • End Stage Liver Disease • Fractures, Bone

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/905807

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Background

Fractures are a common cause for presentation to pediatric A&E in the UK and USA, accounting for up to 14% of admissions. Pre-existing medical conditions and bone disease may also predispose to pathological fractures and produce similar features to CAN fractures [1]. In the presence of a mixed history of child abuse indicators and a pre-existing medical condition, practitioners are faced with the difficult task of balancing the need to maintain their index of suspicion for abuse and preventing further harm with incorrectly diagnosing abuse, which has serious and distressing consequences for both the child and family.

Here, we present a case to illustrate such a dilemma in a child with progressive familial intrahepatic cholestasis (PFIC) type 2 who presented with "red flag" fractures indicative of child abuse. In doing so, we highlight some of the important considerations for the management of suspected non-accidental injury in the context of the pathophysiology of a rare hereditary disorder that can result in pathological fractures.

Case Report

A 22-month-old girl was admitted to A&E after tripping on a carpet and falling to her knees while playing at a local restaurant. Following the fall, she was reluctant to weight-bear on her left leg but she did not look like she was in severe pain.

The child was known to have PFIC type 2 and was listed for liver transplantation. She had been born in good condition at 37+5. Jaundice had been noted on day 5 that persisted to 12 weeks. Further investigations led to a diagnosis of PFIC type 2. She was prescribed fat-soluble vitamins and antihistamines to reduce the itching associated with cholestasis.

The girl underwent venipuncture of her right hand by a pediatrician, at which time a swelling was noted in her hand that was initially thought to be iatrogenic. However, the family also reported that 8 days previously the child had slid down a slide headfirst and used her arms to stop herself at the bottom. Her parents brought her to A&E but the child was well on assessment with no signs of injury, so she was sent home. Documentation of this event was available from the A&E records.

However, in view of possible multiple fractures, including a fractured femur, further investigations were performed and Children's Social Care were notified.

Plain X-rays revealed a non-displaced fracture in the sub-trochanteric region of the left femur (Figure 1A) and an 8- to 10-day-old fracture in the distal right radius (Figure 1B). In view of these multiple fractures, a skeletal survey was performed, which revealed marked osteopenia and widespread metaphyseal abnormalities with widening, splaying, and fraying most evident in the wrists, knees, and ankles (Figure 1C). The survey highlighted the recent fracture of the medial cortex of the proximal left femur with the configuration of a stress fracture and the fracture of the distal right radius with volar angulation of the distal fracture fragment and periosteal reaction, which had increased since the initial plain X-ray. There was also a buckle fracture of the base of the fifth metacarpal without periosteal reaction, perhaps indicating another recent injury.

A bone scan showed increased uptake in the left proximal femur, distal right radius, and right fifth metacarpal consistent with the skeletal survey. There was also a small focus of increased uptake at the 8th left costotransverse junction without a correlate on the chest X-ray.

Her biochemistry revealed calcium 1.70 mmol/L (2.20–2.70 mmol/L), alkaline phosphatase 1728 U/L (60–425 U/L), parathyroid hormone 29.3 pmol/L (1.6–6.9 pmol/L), and vitamin D 18 nmol/L (low). The blood results were entirely consistent with vitamin D deficiency, and the skeletal survey revealed rickets and fractures.

A fractured femur and multiple fractures should always raise the index of suspicion for non-accidental injury and potential CAN. On the surface, the fall in the restaurant did not appear to be of high enough force or velocity to fracture the femur, which usually requires a high-energy accident or trauma in children.

The orthopedic team decided to treat her femoral fracture by immobilization in a hip spica for 4 to 6 weeks. After lengthy multidisciplinary discussions between specialist radiologists and consultant pediatricians and hepatologists involved in the care of the child, a consensus decision was made that the fractures were pathological secondary to hepatic rickets. The child was sent home with her parents. Children's Social Care were made aware of the outcome so they would be able to monitor any changes in the child's or parents' circumstances, although they are no longer involved now that a definitive diagnosis has been made.

Discussion

In this case, although the fractures were ultimately classified as pathological and secondary to hepatic rickets, the team adopted a cautious and guideline-driven approach to the child's management, which included full skeletal survey and referral to Children's Social Care. An alternative approach might have been to postpone informing Children's Social Care until

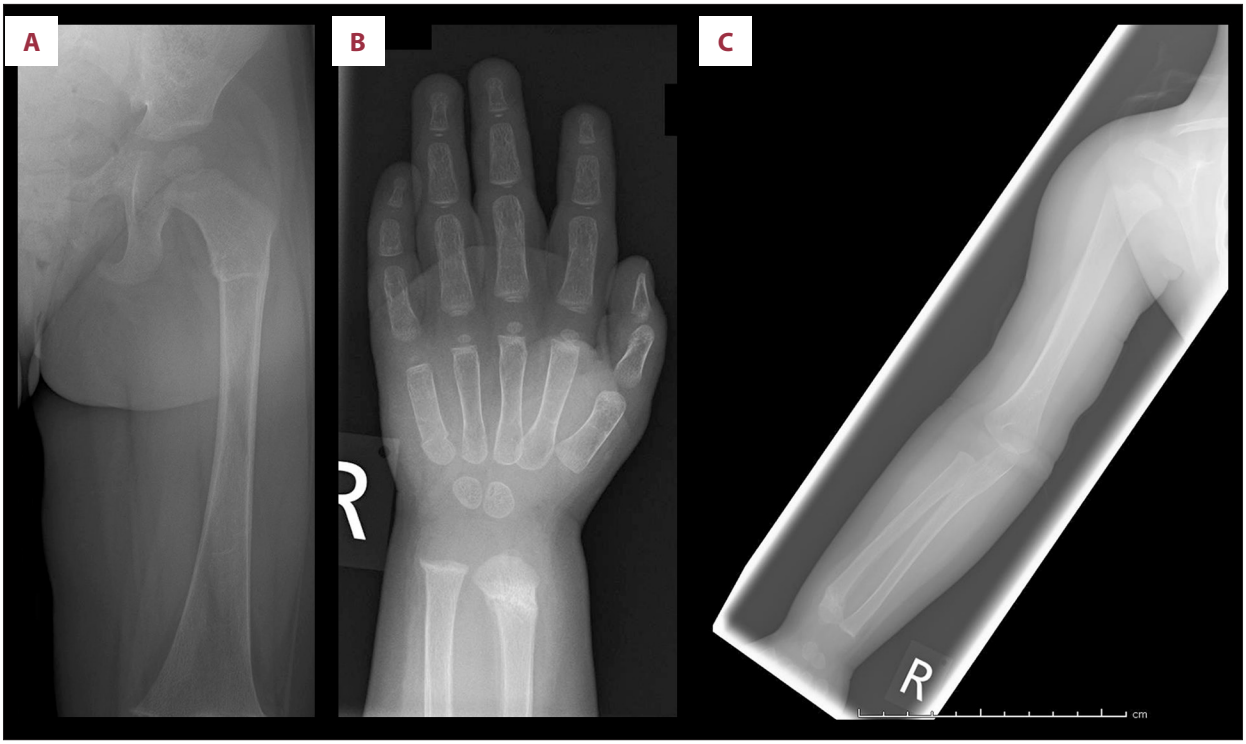

Figure 1. (**A**) Non-displaced fracture, left femur. (**B**) Fracture of the distal right radius with volar angulation of the distal fracture fragment and periosteal reaction. There is also a buckle fracture of the base of the fifth metacarpal but no convincing periosteal reaction. (**C**) Severe osteopenia of the right upper limb.

investigations had been completed, but safeguarding must remain an active consideration when there are circumstances to prompt suspicion of child abuse, and early involvement of other team members can help in decision-making. In this case, external expert assessments were also required to fully conclude that rickets was the most likely cause of the fractures, which introduced some delay.

This case highlights that although it is often difficult to balance different lines of evidence in cases where CAN falls into the differential diagnosis, a pre-existing medical condition does not completely exclude the possibility of non-accidental injury. A safe approach to management of these cases includes a full enquiry of the medical and social history, the age of the child, the location and type of fracture, the mechanism of injury, and excellent communication with other professionals and agencies expert in CAN so that appropriate follow-up can be arranged. In addition, it is imperative that practitioners adopt a sensitive and understanding approach when explaining Children's Social Services referrals to parents, who may understandably become defensive, especially when already dealing with the highly stressful situation of having a sick child.

Progressive familial intrahepatic cholestasis (PFIC), also known as Byler's disease, is an uncommon (1: 50 000 to 1: 100 000 births) autosomal recessive hereditary disorder caused by mutations in transport proteins involved in bile formation [2]. The defective bile transport results in severe intrahepatic cholestasis via different mechanisms, depending on the specific mutation and progressive chronic liver disease, which if left untreated results in end-stage liver disease and death [3]. Our patient showed the classical presentation of PFIC type 2 in the neonatal period, a normal γ-GT and raised AST, and cholestasis with jaundice and pruritus, often the most distressing symptom to the child [3].

Rickets is common in PFIC, with 26/33 PFIC patients showing radiological evidence of rickets in one study [4]. Hepatic rickets occurs primarily due to a failure of metabolism of vitamin D to its active compounds by the damaged liver, but also because patients with PFIC have a disrupted enterohepatic circulation, bile acid deficiency, and possibly reduced circulating albumin and vitamin D-binding proteins secondary to liver dysfunction [5]. Here, the biochemical and radiological findings were consistent with rickets; therefore, this patient was at increased risk of pathological fractures [6].

In this case, there appeared to be a highly plausible medical reason for multiple pathological fractures. Apart from the fractures, there were no other clinical history or physical examination "red flags" such as a history inconsistent with the injury, over 24-hour delay in presentation, or additional injuries

such as bruises [7]. Furthermore, the parents had on 2 occasions promptly sought medical help, which suggests accidental injury, and the parents were compliant and cooperative with all procedures, including Children's Social Care referral, despite this undoubtedly being a difficult process for them.

Nevertheless, child abuse can still occur in children who are unwell. The injuries were not witnessed, and the presence of multiple fractures, including a subtrochanteric femoral fracture, at the very least meant that child abuse remained in the differential diagnosis. There was also a delay in gathering expert opinions from all members of the team at tertiary centers. However, we used Royal College of Radiologists and Royal College of Paediatrics and Child Health Guidelines, also used by the European Society of Paediatric Radiology, and undertook a full skeletal survey since our patient was under 2 years of age [8]. This was an appropriate course of action to identify and document the fractures in case of any future changes to circumstances.

Conclusions

Child protection is a multidisciplinary and multi-agency effort. Particular care must therefore be taken when services are split across sites or jurisdictions since the details of previous contacts with medical services or agencies may not be available. In our case, the child was primarily being managed by pediatric hepatologists in a tertiary center in a different and distant city, so liaison with the family doctor, health visitor, tertiary liver unit team, and social work were necessary to fully inform our decision-making.

Conflict of Interests

None.

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