Unravelling the radiological mystery in a newborn

M. Kueh[1], R. Naffa[2], S. Manickam[2]

1 Queen Alexandra Hospital, United Kingdom, 2 Basildon University Hospital, United Kingdom

Introduction

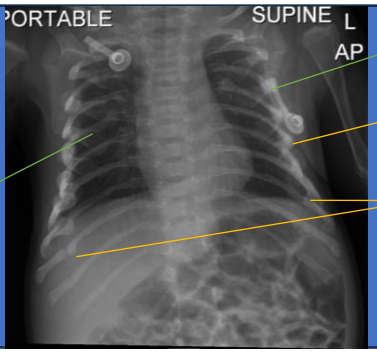
Osteogenesis Imperfecta (OI) is a brittle bone disease which results in frequent and debilitating fractures affecting children and adults. Children who present with these fractures are often mistaken as victims of non-accidental injury and therefore the subject of safeguarding enquiries. Recognition of radiological features of OI or other metabolic bone diseases can prevent undue distress and trauma to the patient and their parents. We present a 20-day old patient with radiological features of Type V OI.

case

20-day old female presented to A&E as her parents were concerned regarding recent weight loss (< 2nd centile), coryzal symptoms, two 30 second apnoeic episodes and two occasions of vomiting. Notably the patient had normal white sclera. (1)

2 investigations were requested:
Chest X-ray ?Pneumonia

MRI head ?Perinatal Hypoxia



(3)

Multiple bilateral healing rib fractures of different ages

Bilateral acute rib fractures (not initially picked up)

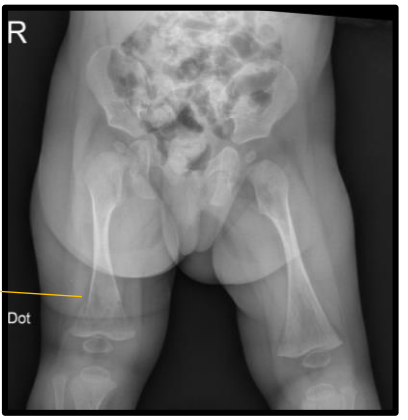
MRI head reported as normal. Patient sent home

6 months later, the child presented to A&E but this time with difficulty in moving her right leg. According to her mother, she was changing her nappy when she felt a snap.

Eccentric periosteal reaction

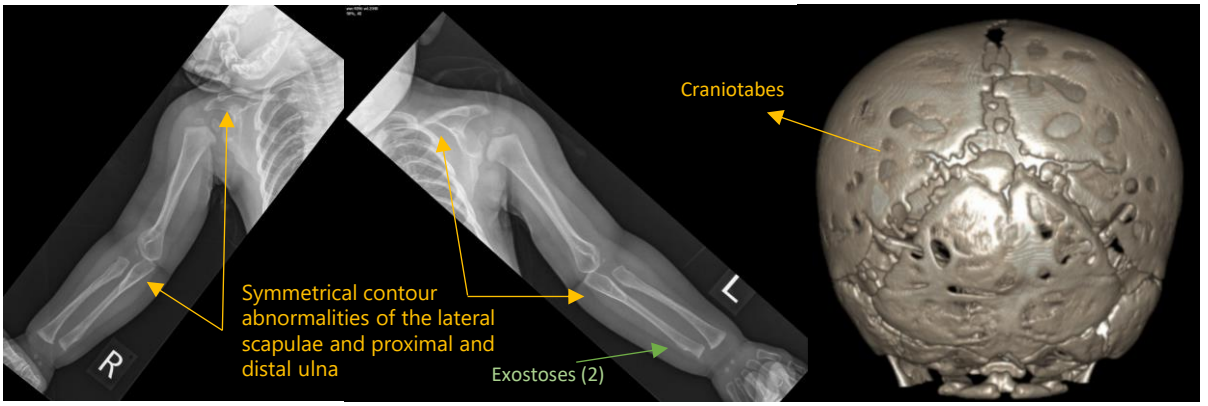
Distal metaphyseal fracture of the right femur

Suspicion of NAI raised; safeguarding measures implemented



Skeletal Survey

Findings raised possibility of metabolic bone disease



Outcome

Patient was referred to the paediatric MDT who determined with the help of radiological findings, that the patient has Type V Osteogenesis Imperfecta. She was started on pamidronate, and further genetic testing was done.

Conclusion

1. Early recognition of metabolic bone disease, in this case features of OI type V can prevent traumatic safeguarding measures to the patient and their guardians.
2. Radiological features of OI V are (1) slender ribs, metaphyseal changes, (2) mineralised interosseous membranes, (3) hyperplastic calluses. Clinically, patients with OI V do not have the classical blue sclera[1].

References: 1 - Kim OH, Jin DK, Kosaki K, Kim JW, Cho SY, Yoo WJ, Choi IH, Nishimura G, Ikegawa S, Cho TJ. Osteogenesis imperfecta type V: clinical and radiographic manifestations in mutation confirmed patients. Am J Med Genet A. 2013 Aug;161A(8):1972-9. doi: 10.1002/ajmg.a.36024. Epub 2013 Jun 26. PMID: 23804581.