Case Report

Metaphyseal and posterior rib fractures in osteogenesis imperfecta: Case report and review of the literature

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ABSTRACT

Purpose: Metaphyseal corner fractures and posterior rib fractures are thought to only occur in settings of inflicted injury. We describe a case of siblings who presented with metaphyseal corner fractures and multiple posterior rib fractures who were later found to carry *FKBP10* mutations, a rare cause of Osteogenesis Imperfecta (OI) known as Bruck syndrome. This clinical presentation led to a literature review examining fracture types in OI and inflicted injury.

Cases: A 15-month-old male presented with multiple healing fractures of varying ages including posterior rib and metaphyseal corner fractures with no history of significant trauma. He had joint laxity, short stature and Wormian bones. His diagnosis of Bruck Syndrome led to investigations in his sibling at birth, which demonstrated the same fracture pattern including multiple posterior rib and metaphyseal corner fractures. They both had pathogenic compound heterozygous *FKBP10* variants.

Literature review and results: We performed a literature review evaluating the fracture pattern in cases investigated for inflicted injury and found to have OI. Fourteen articles reported 78 children with OI initially diagnosed as inflicted injury. Of these children, 71 (91%) were diagnosed with milder forms of OI (Sillence type I and IV). Sixty-four children (81%) had clinical signs of OI including blue sclera, dentinogenesis imperfecta, short stature, joint laxity and limb bowing. Fifteen (19%) children had fractures of high specificity for inflicted injury including metaphyseal corner fractures and posterior rib fractures and 58 (74%) had fractures of moderate specificity for inflicted injury such as bilateral fractures and fractures of different ages.

Conclusion: Metaphyseal corner fractures and posterior rib fractures are highly associated with inflicted injury, but they have been reported in children with OI. Bruck syndrome, a rare and severe form of OI can present with metaphyseal and posterior rib fractures, including at birth. When features of OI are present in children with metaphyseal corner fractures and/or posterior rib fractures are present, genetic testing may be warranted.

1. Introduction

A child presenting with unexplained fractures can be a diagnostic challenge with a differential diagnosis including but not limited to inflicted injuries, Osteogenesis Imperfecta (OI), Menke's syndrome, hypophosphatasia, severe vitamin D deficiency, vitamin C deficiency, and systemic diseases that affect bone metabolism (Flaherty et al., 2014 24470642; Metz et al., 2014 24748639). Posterior rib fractures,

metaphyseal corner fractures and multiple fractures at presentation are thought to be highly specific for inflicted injury in infants due to the mechanisms and shearing forces that would be difficult for an infant to generate (Johnson and Bradshaw, 2015; Kleinman et al., 2015). Unfortunately, inflicted injury is the most common cause of unexplained fractures in infants (Smith, 2000). OI is the most common heritable disorder of bone fragility, but is still rare with a prevalence of one in 15,000–20,000 births (Orioli et al., 1986; Stevenson et al., 2012). While
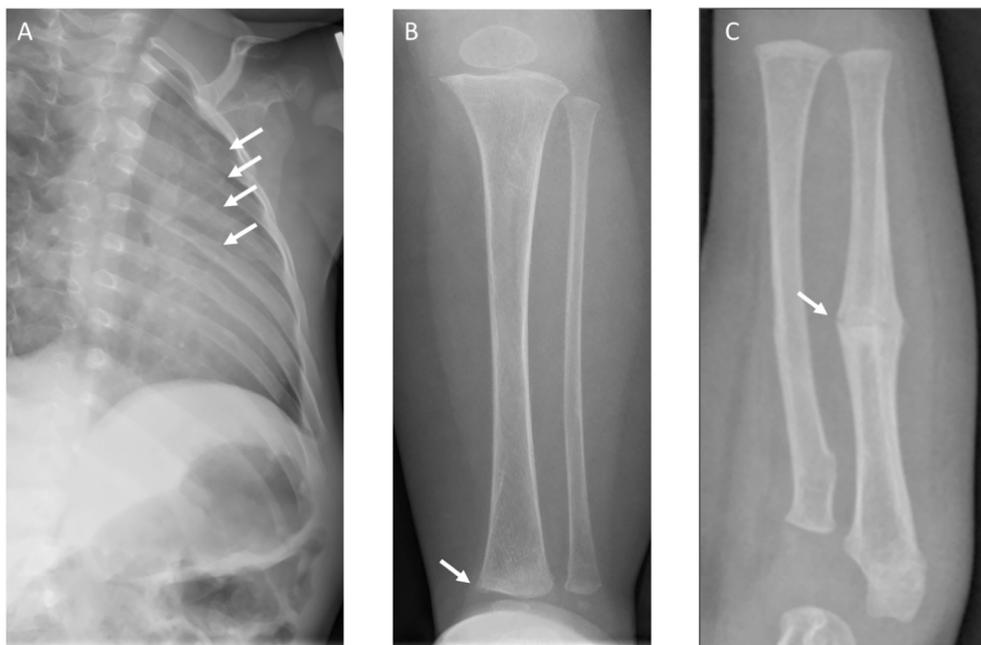

Fig. 1. Skeletal survey findings in 15-month-old infant with compound heterozygous *FKBP10* variants (c.918-3C > G, c.1424_1428dup). A) multiple posterior rib fractures. B) Metaphyseal corner fracture of left medial distal tibia. C) Right mid diaphyseal fracture of the left ulna with callus formation.

some forms of OI present with clinical signs such as blue sclera, dentinogenesis imperfecta (DI) and a clear family history, up to 25% of those diagnosed do not have extra-skeletal manifestations (Pereira, 2015). As a result, OI may be overlooked as a cause of multiple fractures in cases being investigated for inflicted injury (Marlowe et al., 2002; Pepin and Byers, 2015).

Bruck syndrome is a rare autosomal recessive form of OI due to mutations in the *FKBP10* gene which encodes the chaperone protein FKBP65, involved in collagen cross-linking (Alanay et al., 2010; Gjaltema et al., 2016; Lietman et al., 2014). Mutations in *FKBP10* have been shown to cause a clinical spectrum from isolated OI to congenital contractures with severe OI. Effected individuals can have fractures at birth and recurrent long bone fractures can be progressively deforming (Alanay et al., 2010; Shaheen et al., 2011; Steinlein et al., 2011). Wormian bones and joint hyper-flexibility are common findings. Pterygia (skin webbing) and joint contractures may be present at birth or can develop over time (Kelley et al., 2011; Shaheen et al., 2011). There have been reports of rib fractures in two infants with *FKBP10* mutations, although it is unclear where the rib fractures were located and why the infants were investigated (Kelley et al., 2011; Shaheen et al., 2011). No previous report has described metaphyseal fractures in individuals with

OI due to *FKBP10* mutations.

We present the case of a sibling pair with metaphyseal corner fractures and posterior rib fractures, found to have *FKBP10* pathogenic variants causing OI type XI or Bruck syndrome (OMIM 259450). The fracture pattern in these two cases demonstrate that metaphyseal corner fractures and posterior rib fractures can occur in children with OI. This prompted a literature review examining the fracture pattern in children with OI who were investigated for inflicted injury. The hypothesis of this study was that metaphyseal corner fractures and posterior rib fractures can occur in OI and are not exclusive to inflicted injury.

2. Cases

A healthy 15-month-old male was noted to have asymmetric shoulder alignment by his primary care provider. Pregnancy and delivery were unremarkable with no concerns of fractures at birth. Birthweight was 3280 g (Z-score + 0.1) and length 50 cm (Z-score -0.1). He had a history of a left wrist contracture noted at two weeks of age that required splinting. At this time, he was four-point crawling. Family history was negative for OI or recurrent fractures. The parents were healthy, maternal age was 33 and paternal age was 34, and they were non-
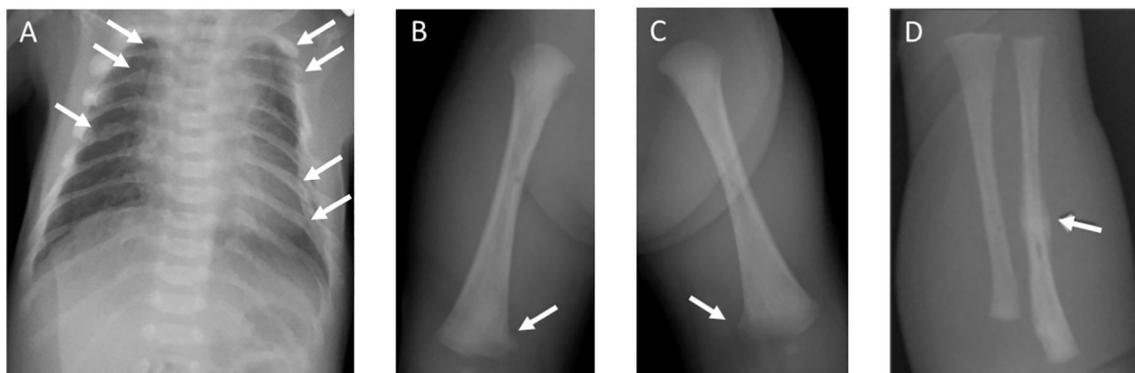

Fig. 2. Skeletal survey findings at birth of sibling with *FKBP10* variants. A) Multiple posterior rib fractures. B&C) Bilateral femoral distal femur metaphyseal corner fractures. D) Left diaphyseal ulnar fracture with callous formation.
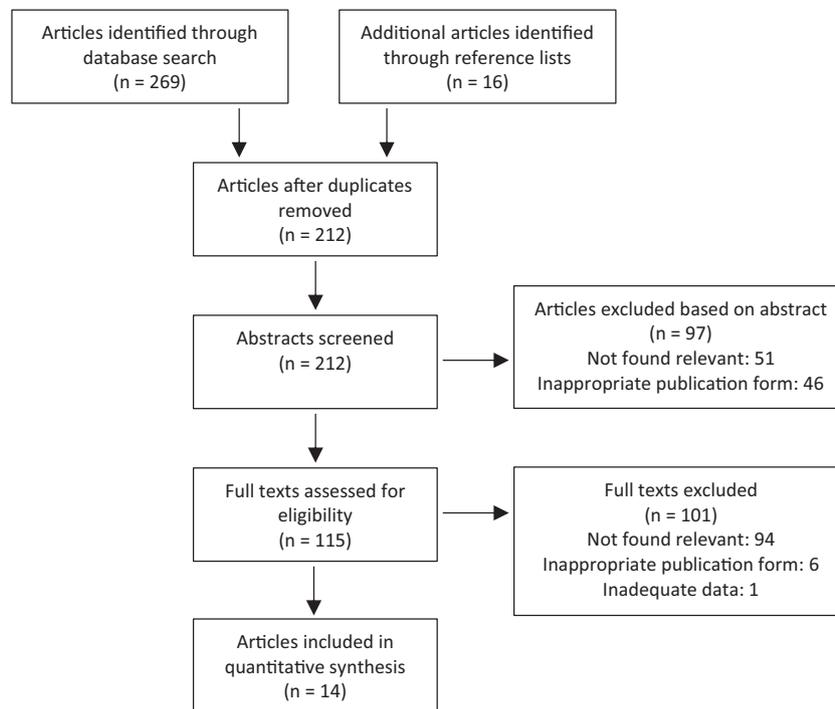
Fig. 3. Literature review process and results.

consanguineous. Physical exam showed short stature (Height *Z*-score -2.0, mid parental height Z-score 0.0), mildly blue sclera, and kyphosis. He had hyperflexibility of his lower limbs including the ability to flex ankles so that his toes touched his shins, pes planus, and hyperflexibility of the hips and knees. There were no findings of dentinogenesis imperfecta (DI) or contractures. Initial X-rays revealed multiple posterior rib fractures with callus formation.

As he had no previous history of fractures and with the concerning potential mechanism of posterior rib fractures, the child maltreatment pediatrics team was consulted, and further investigations were completed. A skeletal survey showed a left tibial metaphyseal corner fracture, multiple posterior rib fractures, and bilateral ulnar fractures with callus formation (Fig. 1). Skull X-ray demonstrated multiple Wormian bones. A CT of the head and ophthalmologic assessment showed no evidence of abusive head trauma. The child maltreatment pediatrics team reviewed the investigations and agreed that OI was the most likely diagnosis based on clinical features including below average stature, joint hyperlaxity, and Wormian bones. Genetic testing revealed pathogenic compound heterozygous *FKBP10* variants (c.918-3C > G, c.1424_1428dup), confirming a diagnosis of Bruck syndrome type I (OMIM 259450), a rare form of OI (OI type XI).

The patient's mother was pregnant at the time of diagnosis. The parents underwent genetic testing and each were heterozygous carriers for one of the *FKBP10* variants. Following genetic counseling, the couple decided not to pursue a prenatal diagnosis. The pregnancy was uncomplicated and long bone fractures were not visualized on fetal ultrasound at 33 weeks gestation. The sibling was born via spontaneous vaginal delivery at 37 weeks with a birth weight of 3345 g (*Z*-score +0.9), length 51.5 cm (Z-score 0.0). Because of the family history of OI, he was handled carefully and he had investigations on day one of life prior to hospital discharge. A skeletal survey demonstrated multiple posterior rib fractures, bilateral metaphyseal corner fractures, and a left ulnar diaphyseal fracture (Fig. 2), a similar fracture pattern to the older sibling. Multiple rib fractures and the ulnar fracture had callus formation, suggesting they occurred in utero and may have occurred after the 33-week fetal ultrasound. Genetic testing demonstrated the same pathogenic *FKBP10* variants and confirmed a diagnosis of Bruck syndrome.

3. Materials and methods

The diagnosis of OI in two siblings who had fractures classically believed to be specific for inflicted injury led to a question of whether these fracture patterns had been reported in other children with OI. A literature search was executed by an expert searcher/health librarian (SC) on the following databases: OVID Medline, OVID EMBASE, and EBSCO CINAHL using controlled vocabulary (e.g.: MeSH, Emtree, etc.) and key words representing the concepts "unexplained fractures" and "osteogenesis imperfecta". Detailed search strategies are available in Appendix 1. Database specific filters were used to limit searches to pediatric subjects. Databases were searched from inception to April 2020. The search was updated February 2021. No other limits were applied.

Results (269) were exported to RefWorks citation management software and an additional 16 articles were retrieved from reference lists. Duplicates (73) were removed. All unique titles and abstracts (212) were screened to determine relevance. Inclusion criteria were: (1) English language, (2) the patients were under the age of 18 years, (3) authors discussed fractures that were initially suspected to be inflicted injury but found to be OI, and (4) the patients were formally diagnosed with OI based on the standard of care at that time. Exclusion criteria were: (1) fractures were not reported as a finding and (2) inflicted injury was not formally investigated. Relevant articles were read in their entirety and eliminated after full text review if inclusion criteria were not met. Fig. 3 shows the screening and exclusion process.

Clinical data collected included the child's age at presentation, type of OI diagnosed, features of OI reported, fracture(s) reported and medical investigations. Fractures were classified into high, moderate and low specificity for inflicted injury using Kleinman's categorization, a system which is widely used by experts in the child maltreatment field (Kleinman et al., 2015). We utilized the updated Sillence diagnostic classification of OI to classify patients identified in our literature review as this classification was used in all included articles (Dijk and Sillence, 2014).

Written informed consent was obtained from the family of the siblings in the case description for publication of their story.

Table 1

Reports of children with OI initially diagnosed with inflicted injury.

Study author and publication year	Number of children with OI reported	Age of presentation	Presenting complaint (n)	Type of OI (n)	Clinical features of OI (n)	Diagnostic method (n)
(Augarten et al., 1993)	1	8 months	Repetitive fractures, swelling, decreased ROM	IV	DI	Clinical diagnosis
(D'Eufemia et al., 2012)	1	20 months	Repetitive fractures	I	Blue sclera, joint laxity, leg bowing	DNA analysis
(Gahagan and Rimsza, 1991)	3	12 months 9 years 16 months	Repetitive fractures (3)	IV (3)	Blue sclera (1) Family Hx (1) Short stature (2)	Skin biopsy (3)
(Lamptey et al., 2009)	1	15 months	Acute leg pain	I	Blue sclera	Skin biopsy
(Marion, 2015)	1	13 days	Acute arm pain, unusual limb position, decreased ROM	IV		Skin biopsy
(Miller and Hangartner, 1999)	1	4 months	Arm pain, decreased ROM	I	Blue sclera	Skin biopsy
(Minnis et al., 1995)	1	3 weeks	Acute bilateral leg pain and swelling	III	Blue sclera	Skin biopsy
(Okpechi et al., 2018)	1	3 weeks	Right arm and leg pain, with unusual position and decreased ROM	Undefined	Blue sclera, leg bowing, short stature	DNA analysis
(Paterson and McAllion, 1989)	15	Mean: 8.7 months (3 weeks - 33 months)	Fracture (15)	I (5) III (1) IV (9)	Blue sclera (1), DI (5), Wormian bones (10), FHx (6)	Clinical diagnosis (15)
(Paterson and McAllion, 2006)	12	Mean 7.3 months (3 weeks - 23 months)	Fracture (12)	I (6) III (1) IV (5)	Blue sclera (9), wormian bones (9), FHx (6)	Clinical diagnosis (7) Skin biopsy (4) DNA analysis (1)
(Kocher and Dichtel, 2011)	33	Mean: 7.1 months (1–23 months)	Pain (14), swelling (7), decreased ROM (5), unusual limb position (2)	I (31) IV (1) undefined (1)	Blue sclera (1), DI (1), joint laxity (1), Wormian bones (1), leg bowing (1), FHx (18)	Skin Biopsy (15) DNA analysis (7) Both method (11)
(Steiner et al., 1996)	6		Fracture (6)	I (2) undefined (2)	Blue sclera (5)	Skin biopsy (6)
(Wardinsky, 1995)	1	10 months	Fracture	IV	Blue sclera, short stature	Skin biopsy
(Wardinsky et al., 1995)	1	9 months	Fracture	IV	Blue sclera, osteopenia	Skin biopsy

DI: dentinogenesis imperfecta.

ROM: range of motion.

(n) = number of children reported.

Table 2

Reported cases of children with OI and metaphyseal fractures and/or posterior rib fractures.

Reference	Age at presentation	Metaphyseal or posterior rib fracture history	Other fracture history	Clinical features of OI	Diagnostic method
Gahagan and Rimsza, 1991	16-Month-old male	Metaphyseal fractures - distal tibia (bilateral) and distal fibula (right)	Long bone fractures of radius and tibia (unilateral)	Older sibling diagnosed with OI	Skin biopsy – qualitative defect in type 1 collagen
Miller and Hangartner, 1999	4-Month-old female	Metaphyseal fractures - proximal humerus (bilateral) and distal femur (left) Multiple posterior rib fractures	Left clavicle fracture	Blue sclera, low bone density on CT bone density measurements,	Skin biopsy – quantitative defect in type 1 collagen
Okpechi et al., 2018	3-Week-old Infant	Posterior rib fractures (8th, 9th rib)	Long bone shaft fractures of L femur, R humerus spiral fracture	Blue sclera. Lower limb bowing and osteopenic appearance on skeletal survey	Genetic testing – heterozygous COL1A1 gene mutation (c.371G > T)
Paterson and McAllion, 2006	1-Month-old male	Metaphyseal: Multiple, unspecified number or location	Femoral spiral fracture (right), clavicular fracture	Blue/grey sclera and wormian bones. The mother had blue sclera and history of multiple fractures in childhood	Skin biopsy – abnormalities in skin biopsy
	3-Month-old infant	Metaphyseal – radius (right)	Long bone fractures of the humerus, tibia and fibula. Multiple rib fractures.	Blue sclera	Skin biopsy*

[*] The original manuscript is unclear about specific means of diagnosis. All attempts were made to contact the author for clarification.

4. Results

The literature review identified 212 articles for screening and fourteen articles met inclusion criteria. They described 78 children who were initially diagnosed with inflicted injuries and subsequently diagnosed with OI (Table 1). The mean age at presentation was 9.3 months

(range 13 days to 9 years) with 23 males (29%), 22 female (28%), and 33 children with unspecified sex (42%). Presenting features included concern of fracture (*N* = 41), unusual limb position (N = 4), decreased limb movement (*N* = 9), swelling (N = 9) and pain (*N* = 19). Twenty-seven (35.1%) of the children had an unclear injury history or unknown cause of fracture.

Fifteen children (19%) had fractures classified as highly specific for inflicted injury including metaphyseal fractures, posterior rib fractures and non-posterior rib fractures (Kleinman et al., 2015). Ten children (13%) had non-posterior rib fractures. Five children (6%) had metaphyseal and/or posterior rib fractures including three children with metaphyseal fracture, one child with posterior rib fracture, and one child with both metaphyseal and posterior rib fractures. These five children all had additional fractures at presentation, and all had features of OI (Table 2). OI was confirmed using skin biopsy and fibroblast culture or gene analysis. See Table 2 for further clinical detail.

There were 58 children (76%) with fractures moderately specific for inflicted injury including multiple fractures at presentation (*N* = 54, 69%), fractures with varying callus formation (*N* = 37, 48%) and bilateral fractures (*N* = 13, 17%). Twenty-nine children (37%) had both multiple fractures and varying callous formation, 10 children (13%) had multiple and bilateral fractures, 1 child (<1%) had bilateral fractures and fractures of varying callous formation, and 3 children (4%) had all three of these moderate specificity features.

Sixty-four (82%) children had clinical features of OI on examination, x-ray or on history (Table 1). 14 (18%) children did not have features of OI, all of whom were diagnosed with a milder form of OI, defined in these cases as Sillence type I or IV. Most children (*n* = 71, 91%) were diagnosed with milder forms of OI based on Sillence classification (Sillence type I or IV), while only 3 children (4%) were found to have more severe forms of OI (Sillence type III). Diagnostic methods to confirm OI were skin biopsy (*N* = 34), DNA analysis (*N* = 10), both skin biopsy and DNA analysis (*N* = 11), and clinical investigation (*N* = 23).

5. Discussion

We report the case of a pair of siblings with Bruck Syndrome, a rare recessive form of OI, who presented with metaphyseal corner fractures and posterior rib fractures, including an infant with fractures at birth. These siblings' cases indicate that fractures thought to be highly specific for inflicted injury can occur in children with OI. Bruck syndrome due to *FKBP10* mutations is a rare form of OI and most individuals will have long bone fractures that are progressively deforming. To our knowledge, metaphyseal corner fractures have not been previously reported in Bruck syndrome.

Our literature review further indicates that fracture patterns classically associated with inflicted injury can occur in children with OI. Most of the children identified had mild forms of OI, and a subset had no clinical features such as blue sclera, dentinogenesis imperfecta (DI), joint laxity, limb bowing, Wormian bones or family history. When specifically examining the reports of metaphyseal and posterior rib fractures in children with OI, it is clear that these are exceedingly rare, but have been reported. Inflicted injury may be misdiagnosed in milder forms of OI because of the milder clinical presentation and/or lack of clinical features (Kocher and Dichtel, 2011). It is important to note that inflicted injury can occur in children with OI or other fragility disorders and a genetic diagnosis of bone fragility does not exclude maltreatment.

Limitations of this literature review include a lack of prospective studies and gaps in data from older studies as far back as 1989. The quality of studies used is important to consider as many were case reports, and several have low quality methods or data presented. Additionally, many articles lacked specific clinical information including locations and numbers of fractures such as location of rib fractures. Older studies diagnosed children with OI based on clinical features such as blue sclera and Wormian bones, both of which can be found in the general population (Brooks, 2018; Marti et al., 2013). Diagnostic ability

for OI has improved significantly over the years, with improved access to genetic testing, thus, the quality of literature had methodological challenges. Nonetheless, the reports of metaphyseal and posterior rib fractures are valuable findings to share with clinicians.

6. Conclusion

These cases add valuable information to the developing knowledge surrounding rare autosomal recessive forms of OI, such as Bruck Syndrome. Our case presentation and literature review consolidate evidence supporting the potential for metaphyseal corner fractures and posterior rib fractures in children with OI and identify these fractures as possible presenting features in Bruck syndrome. If there are features suggestive of OI in children with metaphyseal corner fractures and/or posterior rib fractures, pursuit of genetic testing may be warranted as these fractures are not exclusive to inflicted injury.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.bonr.2022.101171.

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