Hip Dysplasia and Osteogenesis Imperfecta: Case Review and Management Recommendations

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Abstract

A 1-week old female presented with bilateral dislocated hips and was subsequently treated in a Pavlik harness. Harness treatment failed, requiring a closed reduction and spica cast application. In the Post-anesthesia Care Unit, the patient was found to have a right humerus fracture. Six weeks after cast application the patient was found to have nondisplaced bilateral femur fractures, prompting a genetics evaluation. The patient was subsequently found to have osteogenesis imperfecta Type 3. Perioperative fractures in pediatric patients should raise suspicion for osteogenesis imperfecta. Early diagnosis can improve the management of hip dysplasia and allow for early bisphosphonate therapy.

Introduction

Osteogenesis imperfecta (OI) is a group of inherited disorders characterized by brittle bones. The disorder can manifest as easily fractured bones, hearing loss, a curved spine, brittle teeth, or weak muscles (1). The severity of symptoms ranges from mild to severe, and the combination of symptoms can vary. This disorder is commonly associated with pathogenic variants in type 1 collagen that lead to decreased bone quality and quantity (2). The most common pathogenic variants are located in either the *COL1A1* or the *COL1A2* gene, which encode for the alpha chains of collagen type 1 (3).

Hip dysplasia in infants affects 1–3% of newborns (4). Factors such as a positive family history, perinatal breech positioning, and ligamentous laxity put a newborn at increased risk for developing hip dysplasia (4, 5). Testing infants with known OI for hip dysplasia can pose challenges due to the concern for fractures while performing the Barlow or Ortolani maneuvers, or the false negatives that occur with pre-existing femoral deformities (6). This places patients at risk for a delay in diagnosis and future care (7). In subtler OI phenotypes, the diagnosis may not be established at the time of hip dysplasia treatment as management begins early in infancy.

Due to the low frequency of the condition, treatments are poorly described in literature and are often performed under the surgeon's discretion. The purpose of this case report is to describe the presentation and treatment plan of a child born with OI and bilateral hip dysplasia. The patient's family was informed that data concerning her case would be submitted for publication, and they provided consent.

Case Presentation

A female child presented at 1 week of age with bilateral dislocated hips. She was the product of an uncomplicated, term, vaginal delivery. She was the third of four children in the family. All other children were healthy. The patient's initial evaluation demonstrated a right hip dislocation (positive Ortolani) and left hip subluxation (positive Barlow) on exam. She had symmetric hip abduction with a combined abduction angle of 150 degrees with an internal rotation of 60 degrees, and external rotation of 50 degrees. The patient was initially treated with a Pavlik harness. At 5 weeks old the patient had an ultrasound that demonstrated bilateral unstable dislocated hips. At 8 weeks of age, the patient was taken to the operating room for an arthrogram and closed reduction (Figures 1A-C). There were no intraoperative complications; however, in the Post-anesthesia Care Unit (PACU) she was found to have a right humerus fracture (Figure 2A). This was determined to be secondary to positioning at the time of spica cast placement. She was placed into a sling for two-and-a-half weeks (Figure 2B).

Per routine protocol, the patient was taken back to the operating room at 6 weeks after the initial cast placement for bilateral hip examination and application of a new spica cast. This allowed for a skin check and an assessment of hip stability.

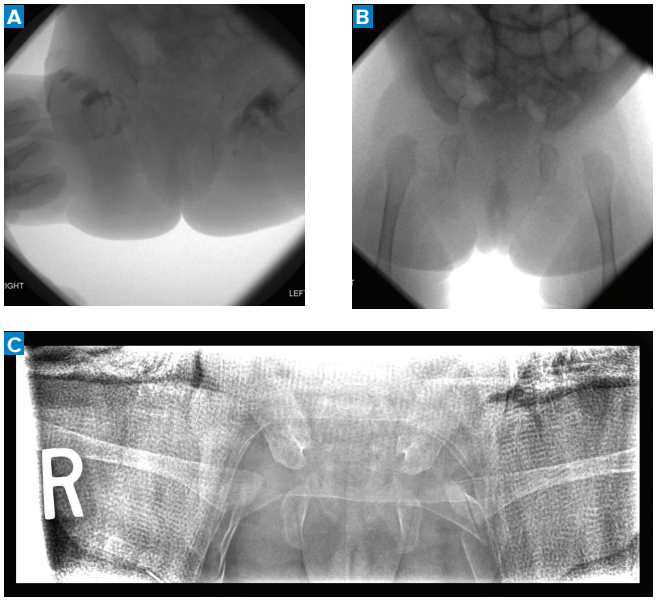

Figure 1. A) Intraoperative arthrogram at 8 weeks demonstrating displacement of both hips. B) Intraoperative bilateral hip arthrograms showing the reduction of the femoral heads into the acetabulum. C) This post-operative X-ray demonstrates the successful relocation of both hips.

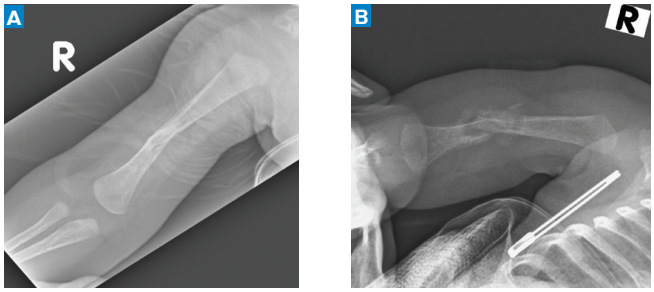

Figure 2. A) This X-ray of the patient at 8 weeks old shows the humerus fracture that was discovered while the patient was in the PACU. B) This X-ray was taken 2 weeks after the identification of the humerus fracture and shows callus formation and periosteal reaction.

The patient's hips were found to be reduced and in the safe zone of 60 degrees (Figure 3). Due to scheduling conflicts, the second cast was removed at 5 weeks instead of 6 weeks. At that time, the patient had concentrically reduced hips with good safe zones. Intraoperatively she was found to have periosteal reaction in both femurs (Figure 4). Rather than placing her in another spica cast, it was decided to place the patient in a Rhino abduction brace for nights only. Approximately 2 months after her second spica removal, she sustained a right proximal femur fracture while being held by her mother and was treated with another spica cast for 6 weeks (Figure 5). The patient was referred to genetics and subsequently diagnosed with OI Type III. At 10 months of age she was started on pamidronate therapy that was continued for 15 cycles over a several-year period. Despite this treatment, she experienced several fractures in her upper and lower extremity that were treated with non-operative management. Follow-up examination over the ensuing 9 years revealed continued acetabular development and no signs of dysplasia (Figure 6 and 7).

Discussion

The first case report describing the association of hip dysplasia and OI was in 1969. Unfortunately, since then, there has been sparse investigation on the topic, with the majority being limited to case reports/series (8). The initial paper reported on two cases of congenital hip dysplasia in male siblings with osteogenesis imperfecta and noted that their mother had blue sclera and joint hypermobility without a diagnosis of OI. Importantly, the paper discussed joint hypermobility as a common etiological factor in hip dysplasia and demonstrated that the mother and both of the patients had joint laxity (8). Fassier et al. retrospectively reviewed 687 patients with OI, 8 hips in 5 patients were diagnosed with hip dysplasia. In 4 of the 5 patients (80%), there was an association with collagen type-1 C pro-peptide variant and the development of hip dysplasia (8, 9). In the same study it was observed that these patients received delayed diagnoses of developmental dysplasia of the hip (DDH) and delayed treatment potentially due to the fear of causing fracture during clinical testing or the difficulty of performing the examination due to hyperlaxity (9). The results of this study also indicated that the incidence of DDH in patients with OI is higher than in the general population (9).

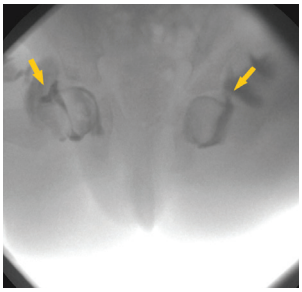

Figure 3. Bilateral hip arthrograms taken at 6 weeks after the first application of the first spica cast demonstrating bilateral congruent hips (rose thorn sign; yellow arrows).

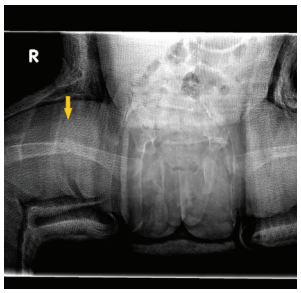

Figure 4. AP Pelvis X-ray conducted after the application of the second spica cast demonstrating bilateral congruent hips and a right proximal femoral deformity (yellow arrow).

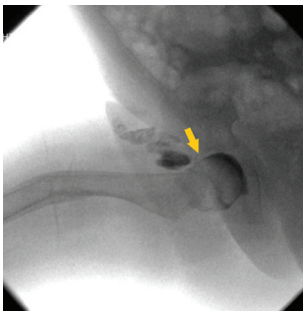

Figure 5. This intraoperative arthrogram was taken at the time of the patient's second spica removal when the decision was made to transition to a Rhino brace. The right hip remains located in this image and periosteal reaction in the femur is noted (rose thorn sign; yellow arrow).

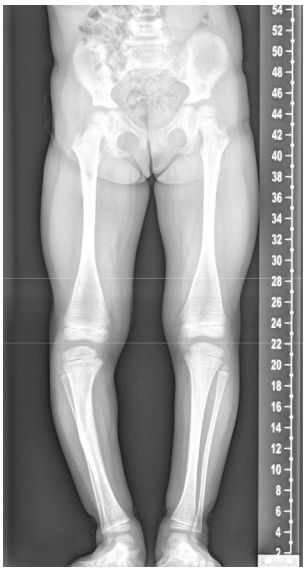

Figure 6. This X-ray of the patient at 4 years old shows that her hips are located and maturing normally.

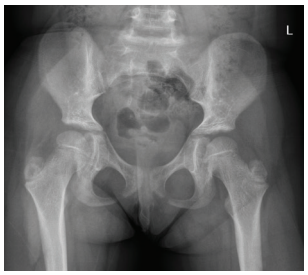

Figure 7. Further follow-up imaging at 8 years of age demonstrating normalized acetabular indices and no significant proximal femoral deformities.

Establishing the diagnosis of DDH in a patient with OI poses additional challenges and risks, as conducting the Barlow and Ortolani screening maneuvers on these patients has been shown to cause femur fractures (6). With regard to our patient's case, it is possible that the femur fracture identified intraoperatively during the second spica cast application may have been caused by these screening maneuvers or manipulation of the child's legs in the operating room, further supporting the previous research showing that physical exam maneuvers may place these children at risk. For this reason, it is suggested that any patient with a family history of OI or clinical features suggestive of OI should undergo alternative methods for evaluation of DDH, such as diagnostic imaging with X-ray or ultrasound (6).

Pavlik harness treatment in the management of DDH has an approximately 70% success rate in young infants (10). In the largest series published, Fassier et al. used a Pavlik harness in two patients (three hips) with hip dysplasia and OI. This treatment was ineffective in both patients and resulted in avascular necrosis in one hip. In our case the Pavlik harness also failed to stabilize the patient's hips, thus requiring closed reduction and spica cast application (11). In addition to the known complications of Pavlik harness treatment, there are also several risk factors that are associated with Pavlik harness treatment failure, which include the severity of the dislocation determined by imaging and age greater than 3 months at the start of treatment (12). However, more likely in our case, the failure of the Pavlik harness could be due to ligamentous laxity or the presence of fractures. This theory is based on studies of children with Marfan's syndrome, in which the researchers found that their generalized joint laxity lead to the failure of Pavlik harness treatment (13). Although not specifically documented in our patient's case, joint hypermobility due to ligamentous laxity is a common feature among several variants of OI. Similarly, Kishta et al. recognized that their patients with OI and DDH also had joint hyperlaxity (9, 13).

Literature surrounding operative management of hip dysplasia in OI shows that treatment goals may differ between age groups. In regard to the older age groups (18 months and older), treatment is generally focused on correcting femoral deformities (9, 14). However, review of the case series of patients who were treated surgically for hip dysplasia in the newborn age group (under 18 months) showed that half were managed with closed reduction and half had an open reduction with or without pelvic osteotomies (9). Reduction was successful after surgical treatment in all but one of these 8 hips (five patients). The open reduction was indicated in children who were older than 18 months. A pelvic osteotomy was necessary in one patient due to significant acetabular dysplasia (9). Our patient was treated with closed reduction and spica cast application, which successfully reduced her hips. In children younger than 18 months without significant acetabular dysplasia, a closed reduction is a successful treatment option for hip dysplasia in OI patients.

Conclusion

Perioperative fractures in pediatric patients, specifically those that occur from positioning in the operating room, should raise suspicion for OI. Similarly, femur fractures that occur while undergoing Pavlik harness treatment are very rare and patients should be referred to genetics for further workup. Prompt diagnosis will help guide management of the hip dysplasia and allow for early discussion of bisphosphonate intervention.

References

1. Devogelaer J-P, Coppin C. Osteogenesis Imperfecta. *Treatments in Endocrinology*. 2006;5(4):229–42.
2. Forlino A, Cabral WA, Barnes AM, Marini JC. New perspectives on osteogenesis imperfecta. *Nature reviews Endocrinology*. 2011;7(9):540–57.
3. Sillence DO, Senn A, Danks DM. Genetic heterogeneity in osteogenesis imperfecta. *Journal of Medical Genetics*. 1979;16(2):101–16.
4. Agarwal A, Gupta N. Risk factors and diagnosis of developmental dysplasia of hip in children. *Journal of Clinical Orthopaedics and Trauma*. 2012;3(1):10–4.
5. Wilkinson J. Persistent Joint Laxity of and the Congenital. *The Journal of Bone and Joint Surgery*. 1964;46-B(1):40–5.
6. Paterson CR, Beal RJ, Dent JA. Osteogenesis imperfecta: fractures of the femur when testing for congenital dislocation of the hip. *BMJ*. 2009;305(6851):464–6.
7. Shorter, D., Hong, T., Osborn, D. A. Screening programmes for developmental dysplasia of the hip in newborn infants. *Cochrane Database of Systematic Reviews*. 2013;54(9):11-54.
8. du Toit S, Weiss C. Congenital dislocation of hips associated with osteogenesis imperfecta in male siblings. A case report. *Bull Hosp Joint Dis*. 1969;30(2):164–70.
9. Kishta W, Abduljabbar FH, Gdalevitch M, Rauch F, Hamdy R, Fassier F. Hip Dysplasia in Children With Osteogenesis Imperfecta. *Journal of Pediatric Orthopaedics*. 2017;37(7):479–83.
10. Ömeroğlu H, Köse N, Akceylan A. Success of Pavlik Harness Treatment Decreases in Patients ≥ 4 Months and in Ultrasonographically Dislocated Hips in Developmental Dysplasia of the Hip. *Clin Orthop Relat Res*. 2016;474(5):1146-1152.
11. Atalar H, Sayli U, Yavuz OY, Uraş I, Dogruel H. Indicators of successful use of the Pavlik harness in infants with developmental dysplasia of the hip. *International Orthopaedics (SICOT)*. 2007 Mar 28;31(2):145–50.
12. Cashman J.P, Round J, Taylor G. Clarke, N.M.P. The natural history of developmental dysplasia of the hip after early supervised treatment in the Pavlik harness: a prospective, longitudinal follow-up. *The Journal of Bone and Joint Surgery*. 2002; 84 (3): 418-425
13. Kerrigan A, Ayeni OR, Kishta W. Developmental Dysplasia of the Hip in Patient with Connective Tissue Disorders. *JBJS Reviews*. 2019; 7(4):e5.
14. Gillingham BL, Sanchez AA, Wenger DR. Pelvic osteotomies for the treatment of hip dysplasia in children and young adults. *JAAOS-Journal of the American Academy of Orthopaedic Surgeons*. 1999 Sep 1;7(5):325-37.