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Arthroplasty in patients with rare conditions

Concomitant achondroplasia and developmental dysplasia of the hip

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ABSTRACT

Achondroplasia (ACH) is the most common form of hereditary dwarfism and presents with multiple musculoskeletal anomalies but is not normally associated with premature hip arthritis. Developmental dysplasia of the hip (DDH) is a spectrum of disease resulting in shallow acetabular depth and a propensity for chronic femoral subluxation or dislocation; it is among the most common causes of premature arthritis. This case report describes the diagnosis of symptomatic DDH in a patient with ACH and highlights difficulties of primary total hip arthroplasty (THA) as a treatment option. Intraoperative radiographic imaging is advised to ensure proper prosthesis placement. Femoral osteotomy may aid visualization, reduction, and avoidance of soft tissue injury. Concomitant ACH and DDH is a challenging problem that can be successfully treated with modified THA.

Introduction

Achondroplasia (ACH) is the most common form of hereditary dwarfism [1,2]. The incidence is estimated to range between 1 and 4.5/30,000 live births [2,3]. Achondroplasia is marked by numerous changes in musculoskeletal anatomy and joint mechanics, but has not been found to be a significant cause of early hip degeneration [4]. Features commonly associated with ACH include a long and narrow trunk, shortened extremities, particularly along the proximal (rhizomelic) segments, bowing of the tibia, and deepening and flattening of the acetabulum [5–7]. Joint laxity is common, most frequently in the knee and shoulder, and manifests as progressive hyperextension of the knees and chronic inferior dislocation of the shoulder, respectively [8,9].

Developmental dysplasia of the hip (DDH) is also characterized by aberrant joint laxity, encompassing a spectrum of pathology culminating in shallow acetabular depth and a propensity for hip subluxation or dislocation. The incidence of DDH is estimated to range between 1 and 5 per 1000 live births [10,11]. DDH is among the leading causes of premature hip osteoarthritis [12]. The hip-specific features of DDH are summarized by Crowe [13,14] and Hartofilakidis [15] (Table 1). Crowe described the range of subluxation of the femoral head as extending from less than 50% (group I) to greater than 100% (group IV) [13,14]. Hartofilakidis described three types of DDH, ranging from only mild acetabular dysplasia to a severe dysplasia of the acetabulum and chronic dislocation of the hip with a pseudo-articulation between the femur and a hollow in the iliac wing [15].

Concomitant DDH in the patient with ACH is a rare occurrence. Here, we present a case of a patient with ACH undergoing total hip arthroplasty (THA) due to osteoarthritis secondary to Crowe IV DDH.

Case history

A 36-year-old female with ACH and bilateral DDH was referred for evaluation due to severe pain and discomfort in both hips (Fig. 1). The pain had increased in severity for the past five months and was greatly exacerbated by activity. As a result of her debilitating pain, the patient was unable to walk a distance of greater than one block. She had tried anti-inflammatory medications, ambulatory assistive devices, and low-impact exercises, for more than 3 months, none of which had provided substantial pain

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Table 1
Crowe and Hartofilakidis staging of developmental dysplasia of the hips.

Classification	Description
Crowe [13,14]	
I	Femoral head subluxation <50% or proximal displacement <10%[a]
II	Femoral head subluxation between 50% and 75% or proximal displacement between 10% and 15%[a]
III	Femoral head subluxation between 75% and 100% or proximal displacement between 15% and 20%[a]
IV	Femoral head subluxation >100% or proximal displacement >20%[a]
Hartofilakidis [15]	
Dysplasia	Femoral head exhibits chronic subluxation but located within true acetabulum
Low disarticulation	Femoral head articulates with false acetabulum partially covering the true acetabulum. Inferior of false acetabulum overlies superior lip of true acetabulum
High disarticulation	Femoral head articulates with a hollow of the iliac wing superior and posterior to true acetabulum. No direct contact between false and true acetabulum

[a] Proximal displacement calculated as distance between medial femoral head–neck junction and the inferior margin of the acetabulum divided by the height of the pelvis.

improvement. The patient's medical history was significant only for ACH and bilateral DDH. Both her father and son have ACH; there was no history of DDH among first-degree relatives reported.

On examination, the patient measured 122 cm (4 ft, 0 in) in height. She had an intact neuromuscular examination of bilateral lower extremities. Examination of both hips demonstrated full passive range of motion. There was significant pain in both hips upon passive internal and external rotation as well as with flexion greater than 90°. Radiographic evaluation demonstrated chronically dislocated hips, with severe degenerative changes about the hip joints bilaterally and pseudo-articulation with the iliac crests consistent with a Crowe IV classification (Fig. 1).

Treatment options were discussed with the patient, with the recommendation being bilateral total hip arthroplasty (THA) in a staged manner starting with the left hip, as it was the more symptomatic side. Intraoperatively, the patient was placed in the right lateral decubitus position and a posterior approach utilized. The pseudocapsule was identified and tagged. The location of the under developed native acetabulum was identified visually, verified with intraoperative imaging, and reamed to accept a 40 mm acetabular cup (Fig. 2). A 40 mm cup was impacted into place with 45° of abduction and 20° anteversion. Two screws were placed to secure the cup. A 22 mm highly cross-linked polyethylene liner was used. The proximal femur was identified and the femoral canal established and reamed to 11 mm. A 4 cm segment of the proximal femur was removed via a shortening subtrochanteric osteotomy to enable reduction (Fig. 3). Prophylactic cables were paced both on the proximal and distal segments to prevent fracture propagation. A modular 11 mm stem was impacted into place and a 22 mm head was used. The resected proximal femur was halved and used as a sleeve over the osteotomy site and held with a 2 mm cable. After reduction, the hip was found to be stable in all planes.

The patient was admitted on the standard total joint replacement pathway and discharged on the second postoperative day with instructions to be partial weight bearing. The first clinic visit was at two weeks after surgery. By that time, the patient was participating well with physical therapy. Radiologic and clinical examination confirmed a stable left hip with no evidence of infection, loosening, periprosthetic fracture, subluxation, or dislocation (Fig. 4a and b). She was kept partial weight bearing on the left hip. At six weeks, the patient was still participating in physical therapy without difficulty or complication; she was instructed to begin weight bearing as tolerated and received a temporary lift for her right shoe to correct the 2 cm leg length discrepancy resulting from left hip restoration. At three months, the patient was ambulating without difficulty and had no complaints at the left hip or thigh; all components were well seated, without evidence of loosening, fracture, or wear. At this time, the patient was scheduled for right primary THA, which was subsequently completed without any complications with a similar protocol (Fig. 5).

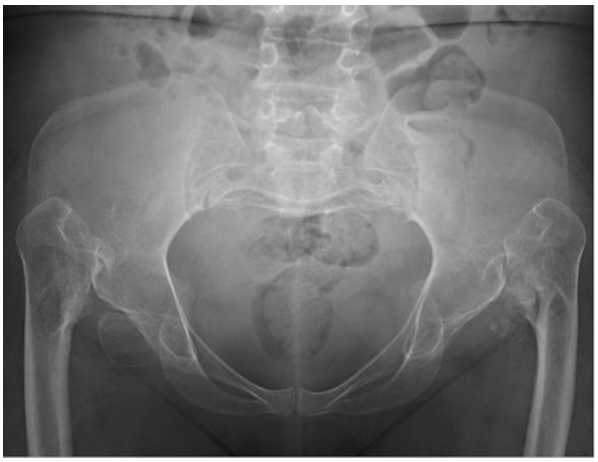

Figure 1. Anteroposterior pelvis view of 36-year-old female with achondroplasia and high hip dislocation, Crowe group IV developmental dysplasia of the hip.

Discussion

In this case report we describe a patient concurrently affected by two musculoskeletal disorders with significantly different pathogenesis.

Achondroplasia

Achondroplasia results from a fully penetrant, autosomal dominant disorder of fibroblast growth factor receptor-3 (FGFR3), phenotypically characterized by sequela of deficient endochondral ossification [16]. More than 95% of cases are thought to stem from a gain of function Gly380Arg amino acid substitution in the transmembrane domain of FGFR3 [16]. FGFR3 has been shown to inhibit chondrocyte differentiation and proliferation largely through its effects on MAPK and Stat1 signaling pathways, respectively [17–19]. Effects of chondrocyte inhibition via FGFR3 are enacted at the physeal plate which are characteristically delayed in maturation and thin until at least the age of puberty [20,21].

Anatomic features of ACH persisting into adulthood include rhizomelic short stature, frontal bossing of the skull, spinal stenosis, pronounced lumbosacral lordosis, shortened lumbar pedicles, flared and splayed metaphysis, and joint laxity [5,22]. Pelvis- and hip-specific characteristics of ACH include a shortened and broadened pelvic cavity leading to a “champagne-glass” pelvis, squaring of the iliac wings, a flattened acetabular roof, a decreased acetabular beta-angle, increased acetabular coverage of the femoral head, genu varum, and a short and broad femoral neck [6,7].