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CASE REPORT

Unpredictability of hip behavior in Dyggve-Melchior-Clausen syndrome: A mid-term assessment of siblings

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Summary Dyggve-Melchior-Clausen syndrome is a rare spondylo-epiphyseal disease, which almost constantly leads to both bilateral hip degeneration and dislocation. Few authors have reported to date the surgical management of this orthopaedic disorder. We present two new cases affecting siblings. One brother was treated by unilateral triple pelvic osteotomy combined with varus osteotomy of the proximal femur; the other was treated by bilateral Pemberton osteotomies with varus osteotomy of the proximal femur. At a respective 5-year and 3-year follow-up delay, both cases had evolved towards progressive subluxation recurrence along with severe hip degeneration. Based on both our experience and literature review, it seems that one should avoid operating these hips unless pain renders surgery mandatory. Total hip arthroplasty seems the only reliable surgical solution at the adult age and paediatric surgeons should keep in mind that previous femoral osteotomies will make it more challenging for adult orthopaedic surgeons to implant on a remodeled anatomy.

Introduction

Dyggve Melchior Clausen (DMC) syndrome is a rare recessive autosomic genetic disease, which mainly affects the skeleton as a spondylo-epiphyseal disease. It was first described in 1962 [1], and since then as few as a hundred cases have been published to date, which accounts for a mean prevalence of 0.1 per million [2]. DMC syndrome is characterized by multiple orthopedic issues, progressive harmonious dwarfism with

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a short trunk and light to severe intellectual impairment [3—7]. We report two new cases of two siblings presenting with a typical DMC phenotype. Progressive hip degeneration and subluxation have already been described in the literature [8], in both operated and non-operated hips in such patients. Few authors have reported their experience in addressing hip subluxation in these children. It seems that whatever the surgical technique chosen to address this hip condition, spontaneous evolution tends towards subsequent subluxation and degeneration of the joint. Our cases moreover present progressive articular fusion in non-operated joints such as the knee, shoulder, wrist and proximal and distal interphalangeal joints.

Observations

Case 1

Case 1 was referred to our institution at age 9, having already been diagnosed with DMC syndrome. He presented with swollen and painful wrists and progressive bilateral hip subluxation. Biological tests ruled out any rheumatologic or infectious causes to the carpal swelling. Subluxation of the right hip eventually became painful at age 17 and a varisation of the upper femur with associated triple pelvic osteotomy were performed. Postoperative course was uneventful and short-term quality of life was improved. However, at 5-year follow-up, both hips progressively evolved towards progressive degeneration with major arthrosis. The right hip is still moderately painful and the femoral head evolved towards subtotal avascular necrosis. The left non-operated hip developed spontaneous coxo-femoral fusion with no possible range of motion (Fig. 1). The carpal joints have remained painful and swollen and the capitate bone progressively showed radiographic signs of articular degeneration.

Case 2

Case 2 is the younger brother of case 1. He was referred to our institution at age 4. Skeletal assessment showed the classical and pathognomonic signs of DMC syndrome but there were no hip subluxation and no clinical symptoms of any kind to report. At age 9 the patient developed subluxation of both hips, but only the right hip was painful. At age 11, both hips had a 75% Reimers index [8] and the patient had great difficulties for everyday activities. Bilateral femoral varisation with associated Pemberton osteotomies were performed. Postoperative course was uneventful but over a 3-year course, the patient rapidly developed progressive articular degeneration with subsequent subluxation of the right hip (Fig. 2). The left hip presents with normal range

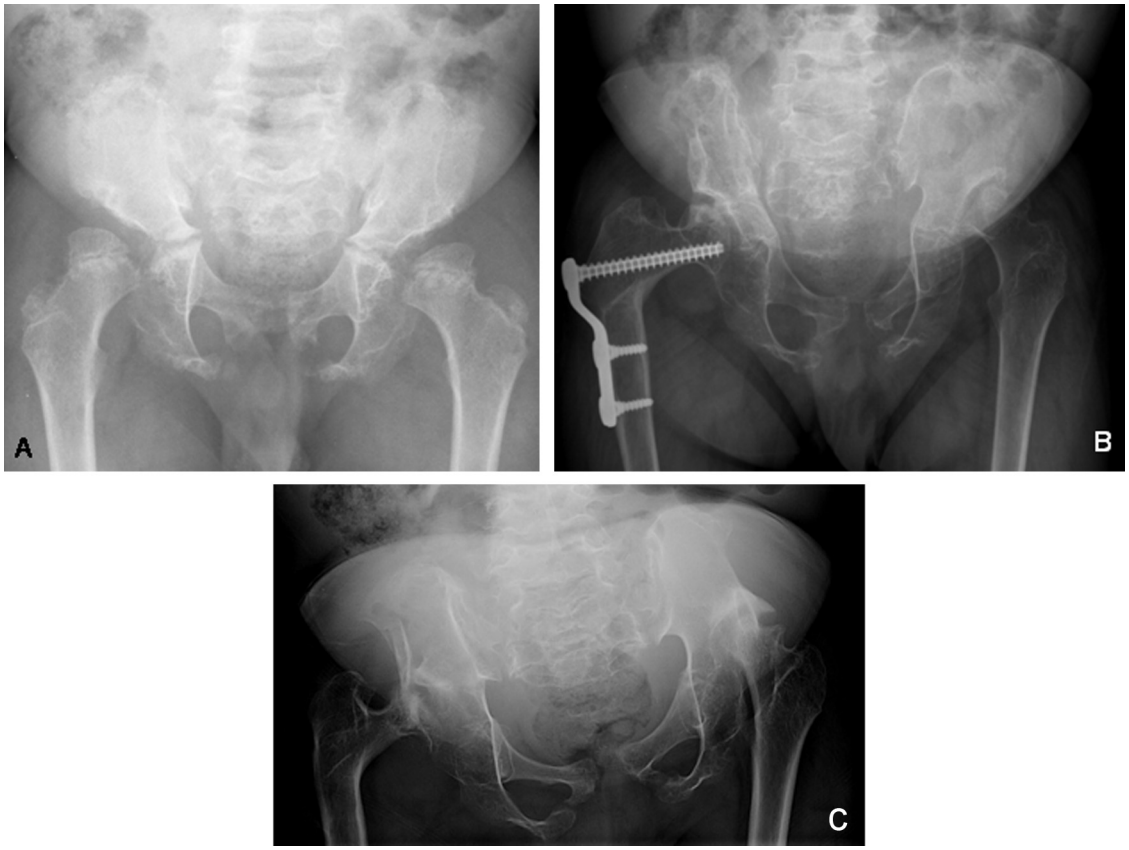

Figure 1 Case 1. A. Preoperative anteroposterior (AP) pelvic view. Note the hypoplastic acetabuli and subluxation of both hips. B. AP view at 1-year follow-up after femoral varisation and triple pelvic osteotomy. The right hip is congruent, but metaphysoepiphyseal osteosclerosis is beginning to appear. Left hip is already deformed with progressive joint disappearance. C. AP view at 5-year follow-up. Mild subluxation of the right hip with an almost complete destruction of the coxo-femoral joints.

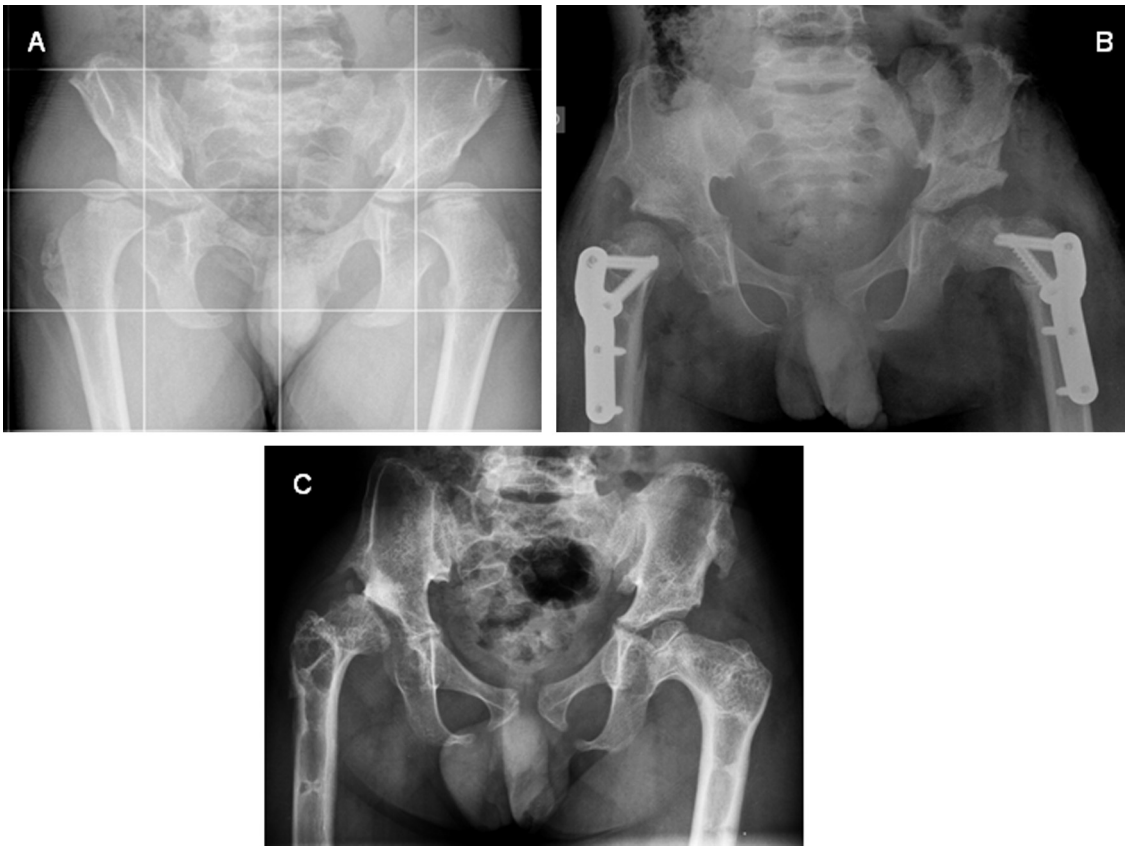

Figure 2 Case 2. A. Preoperative anteroposterior (AP) pelvic view. Note the bilateral subluxation of the hips and hypoplastic acetabuli. B. Postoperative AP view of the hips at 6 weeks follow-up after Pemberton osteotomies and upper femoral varisation. Both hips are adequately congruent and there is no noticeable joint degeneration. C. AP pelvic view at 3-year follow-up. Note the recurrent subluxation of the right hip, while the left hip remains congruent.

of motion and is not painful. He also developed bilateral carpal swelling and right scapulo-humeral swelling with painful active mobilization of the shoulder. Radiographic and scanographic assessment of the wrists show spontaneous perilunate posterior type II dislocation with moderate osteosclerosis.

Discussion

DMC syndrome is a rare spondyloepimetaphyseal disease caused by mutations in a gene mapped to chromosome 18q21.1, coding for a protein called dymeclin, mainly found in fetal cartilage, bone and brain cells [9]. Numerous orthopaedic issues have been described to date in DMC syndrome. Main orthopedic complications include spondylo-epiphyseal dysplasia with lumbar hyperlordosis, scoliosis, thoracic hyperkyphosis, barrel-chest deformities, progressive hip dislocation, knee deformities and medullar compression due to atlanto-axoideal instability [3—7]. Orthopedic hip issues resemble that of patients affected with mucopolysaccharidosis, mainly type IV Morquio's disease, but the latter almost never present mental retardation unlike DMC patients [6]. Thus patients with DMC syndrome often present with a severe chondrodysplasia, important dwarfism, limited autonomy and fairly severe mental retardation. Radiological findings at the hip in DMC syndrome reveal almost constantly a hypoplastic acetabulum, a delayed hypoplastic deformed femoral head and neck and bilateral coxa valga, leading to progressive bilateral dislocation of the hip joint laterally and superiorly [7]. One can also see the almost pathognomonical irregular ''lacy'' iliac crests which help to diagnose the affection [6,7].

Several techniques have been reported in the literature, none of which have been successful at latest follow-up. Hosny and Fabry [4] report a 14-year old girl with DMC syndrome presenting with bilateral hip subluxation. Bilateral Chiari osteotomies were performed but the patient developed subsequent bilateral subluxation at 9-year follow-up. The authors concluded that Chiari osteotomy was not the appropriate treatment.

In our first patient, combined femoral varisation and triple pelvic osteotomy were performed on the right hip. However, functional outcome at 5-year follow-up for both the operated and the non-operated hip was very poor. Despite the initially satisfying radiological outcome, both hips evolved almost symmetrically towards progressive subluxation and upper femur avascular necrosis. Scannographic assessment shows complete disappearance of the joint on the left non-operated hip.

More often are these children referred to orthopaedic surgeons during the first decade. Burns et al. [10]

report seven cases, including one patient with a Sugioka transtrochanteric rotational osteotomy which further required total hip arthroplasty. In case 2, addressing acetabular hypoplasia was our main goal. Since previous surgical procedures in his older brother but also in the literature had all proved failures, it was thought that perhaps the excentricity of the upper femoral epiphysis and the altered morphology of the Y-shaped cartilage [11] had to be taken more accurately into account to prevent such transitory effectiveness of surgical interventions. Therefore were performed bilateral Pemberton pelvic osteotomies combined with upper femur varisation. At three year follow-up however, the right hip presents with progressive subluxation and femoral head avascular necrosis along with painful limitation of range of motion. To date, the left hip remains fairly mobile with no subluxation.

It seems that hip subluxation in DMC syndrome evolves towards complete hip degeneration regardless of the surgical option chosen to address subluxation and prevent further articular lesions. Metaphysoepiphyseal changes in the upper femur are constant observations in the literature [2—4,7,8,12]. It is not well known whether they are responsible for the avascular necrosis of the epiphysis. However such phenomenons are observed as well in other parts of the skeleton. In case 2, scannographic assessment of the left wrist showed perilunate posterior type II dislocation with moderate osteosclerosis. Bone structure was again abnormal with signs of necrosis, osteosclerosis and spontaneous joint dislocation. The lunate bone was hypoplastic. This spontaneous phenomenon is very interesting because it might help to explain the physiopathology of hip dislocation as well. Growth changes, necrosis, bone deformation and hypoplasia, lead in both joints to global joint loosening, with progressive joint hypermobility. Besides MRI findings regarding the growth plates [11], another reason for surgical failure might as well be the global hypoplasia of bone structures around the joints, aggravated because of the additional osteonecrosis leading to bone deformation. Because of this physiopathological continuum, it seems, according to both our experience and the literature, that whatever the surgical option chosen to address hip dislocation, growth changes will inevitably lead to progressive recurrence of the subluxation. In this regard, perhaps should surgical procedures be performed only in case of severe pain in children and adolescents with DMC syndrome. Moreover, clinical features such as limited autonomy and severe mental retardation should discourage aggressive surgical procedures. Thus total hip arthroplasty becomes the only definitive and reliable option at the adult age. Moreover, since it is known that upper femoral osteotomies render femoral stem implantation more difficult [13], perhaps should such osteotomies be avoided in this regard. Both anatomical and mechanical conditions are favorable in these patients, and combined with limited autonomy and mobility, hip arthroplasties should last a very long time.

Conclusion

It seems mandatory when dealing with Dyggve Melchior Clausen to make sure it is the correct diagnosis. One should rely solely on clinical pain and on both surgical literature guidelines and Constitutional Bone Disease Reference Center advices, never on radiological findings, when proposing a surgical solution to address hip subluxation. Proximal femoral osteotomies should be avoided to prevent rendering hip arthroplasty difficult at the adult age. The latter solution seems the most reasonable with regard to growth anomalies, local anatomy, limited autonomy and intellectual impairment in these young patients.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

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