Clinical Case Study

Coxofemoral Instability and Patellofemoral Instability in Down Syndrome

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Abstract

Trisomy of the 21st chromosome, or Down syndrome (DS), is the most common chromosomal abnormality. It is associated with certain musculoskeletal conditions that are usually present to some degree in most individuals with Down syndrome. Musculoskeletal conditions linked to generalized ligament laxity are among the main features of this syndrome. Laxity may sometimes be concomitant with significant joint pathology, such as hip instability or patellofemoral instability. The authors present two clinical cases, one with non-traumatic hip dislocation treated conservatively with a spica cast and prolonged immobilization, and another with patellofemoral instability treated surgically. After 18 and 15 months' follow-up, neither patient has experienced a recurrence.

Keywords: Orthopedic disorders. Ligament laxity. Hip dislocation. Patellar dislocation. Down syndrome.

Introduction

Progress in preventing and treating Down syndrome (DS)-related conditions in recent decades has contributed to the full social integration of this population segment. Psychosocial, educational and legal measures to ensure a full everyday life must be balanced with medical care for organic conditions typically associated with this syndrome.

These include skeletal disorders -- short neck, chest abnormalities, a single palmar fold, a single flexion furrow of the fifth finger, a wide space between the large toe and second toe ("sandal gap") -- which are characteristic, as well as musculoskeletal joint disorders, generally in the form of instability, which may be secondary to a different tissue collagen structure.

Our aim is to focus on describing two types of lower-limb instability that often require medical care. Though not life-threatening, hip instability and patellofemoral instability impair social integration and social activity in individuals with DS, who need early and ongoing stimulation at every level.

Hip instability

Coxofemoral instability is characterized by potentially recurring dislocations; unlike congenital hip dislocation, its underlying cause is not hip dysplasia.

Although the classic DS pelvic morphotype shown by Shaw and Beals (1), with enlarged iliac alae and horizontal acetabulum, should theoretically lead to a
self-stabilizing coxofemoral joint, the wide range of movement due to ligament laxity explains the clunk found upon examination as well as voluntary hip dislocation through contraction of the gluteus muscles. When this coexists with coxa valga and femoral anteversion, the development of a clinical condition is more likely.

Frequent dislocations may affect the growth of the femoral head in two ways: deterioration will be faster, and the femoral neck is more likely to develop a valgus deformity that will increase with bipedalism and lead to progressive subluxation, tending to cause unilateral or bilateral chronic dislocation which becomes very hard to address.

One out of twenty children with DS are thought to develop recurring dislocations between the development and onset of walking and the age of 10 (2). After age 24 months, habitual dislocation requires treatment to prevent dysplasia, subluxation or locked dislocation.

Natural history

Generally, walking is delayed until age 2, and hips are hypermobile during this period. Morphologically, the acetabulum is very deep. Congenital dislocation is rarely found together with this condition, but whenever present, it responds well to conventional treatment.

From 24 months with the onset of walking up to age 10, the hip will spontaneously dislocate in certain positions that the patient can self-reduce. These symptoms are often a cause for seeking care.

After age 7-8, the hip may be in clinical subluxation with moderately painful symptoms. Gradually, subluxation will induce acetabular dysplasia. Without treatment, the natural history of this condition may lead to an irreducible locked dislocation in the second decade. Patients remain able to walk, with moderate pain occasionally requiring anesthesia. At this stage, imaging will show an irreducible iliac dislocation with acetabular dysplasia and false acetabulum (3).

Recurring dislocations appear in patients aged 2 to 10 with considerable ligament laxity. Acute dislocations develop at an age in which laxity has disappeared, well into the second decade of life. Progressive subluxation develops over the second decade and the beginning of the third, until it becomes a locked dislocation.

Currently, surgery for recurring dislocations is the treatment of choice unless anesthesia is contraindicated. Acute dislocation may be reduced and immobilized with a spica cast (4, 5) but surgical treatment is recommended if orthopedic treatment has failed and dislocation has recurred. Highly advanced subluxations and locked dislocations may be treated conservatively; surgical outcomes are considered poor at this stage.

The surgical treatment of choice combines capsule repair and pelvic or femoral osteotomy: varus derotational osteotomy for the femur (6), and acetabular redirection osteotomy or acetabular dome construction (7, 8, 9, 10). The best outcome is achieved by treating patients at the recurring dislocation stage, before the onset of acetabular dysplasia.

Case report 1

A female patient with DS aged 11 years and 7 months, with deep cognitive impairment, presented at the emergency service with abnormal irritability and refusal to rest her weight on her lower limb when getting up from bed. The family stated that she had not been involved in any trauma.

Clinical examination found typical Down syndrome musculoskeletal traits, with significant hyperlaxity. A significant finding was lower limb asymmetry, with a shorter left leg slightly adducted, flexed knee and internal rotation.

An AP x-ray of the pelvis showed a dislocated left hip (Fig. 1).

When asked, the patient’s family reported no prior instance of spontaneous dislocation, so conservative treatment with an immobilizing cast was attempted. To spare the patient some pain and discomfort, the dislocation was reduced under general anesthesia and the hip was found to be significantly unstable, secondary to joint laxity. A spica cast was applied (Fig. 2). Immobilization was maintained for eight weeks and then removed, with the patient allowed to walk. Over the following months, the patient had regular clinical and radiological check-ups, with no findings of impairment secondary to dislocation.

After 18 months’ follow-up, the patient remains asymptomatic and no other spontaneous hip dislocations have developed (Fig. 3).

Patellofemoral instability

Patellofemoral instability is often overlooked in routine pediatric examinations. Reported frequency varies, with different series ranging from 3.3% to 37%. On the whole, most studies report incidence rates between 3% and 20% (11).

Generally speaking, patellofemoral subluxation presents with a tolerable degree of pain that allows the patient to proceed with everyday activities (12). Currently, social integration and integrated sports for
people with DS (13) increase patellar overuse symptoms resulting from minimal patellar misalignment features. This exposes conditions that had previously remained subclinical, some of which can be addressed by using external orthoses for sports (14).

Clinically, patellofemoral instability can be classified as follows: grade I: stable patellofemoral joint; grade II: patellofemoral instability allowing lateral subluxation of the patella greater than 50% without dislocation; grade III, dislocatable patella; grade IV, manually reducible patellar dislocation; grade V, irreducible patellar dislocation.

Such hypermobile patellae tend to have chondromalacia of the patellar cartilage.

Instability occurs with the maximum degrees of hypermobility. Some predisposing factors for patellar dislocation include muscle hypotonia, ligament hyperlaxity, valgus knees, trochlear dysplasia, and changes in the anterior tibial tuberosity and patellar tilt.

Flexion and extension movements readily show the displacement of the patella over the lateral condylar facet. AP and lateral X-rays (15), as well as computerized axial tomography (CAT) to assess patellar tilt and distal alignment, will provide guidance as to the kind of correction required. With open physes, surgical recentering will act on soft tissue without osteotomizing the anterior tibial tuberosity.

While some authors, e.g. Matsusue et al. (14), highlight the importance of considering certain medical aspects such as intelligence, daily activity rate and obesity in treating patellar dislocation in DS, it is important to bear in mind that failure to address recurring patellar dislocations will predispose the patient to permanent dislocation, with an incompetent extensor apparatus which goes into abduction and causes knee flexion and lateral rotation of the tibia. Failure to correct this early will lead to trochlear dysplasia (16); once this is in place, it will be the cause of many recurrences secondary to delayed recentering episodes.
Case report 2

Male patient aged 9 years and 8 months, with DS and slight cognitive impairment, presented with chronic knee pains and repeated falls.

Examination found a bilateral patellar clunk in flexion-extension, with a bilateral patellar dislocation that was refractory to rehabilitation (Fig. 4).

Given the clinical findings, plain X-rays and a CT scan were requested, which corroborated the suspected diagnosis showing significant patellar lateralization (Fig. 5 and 6).

Elective surgery was advised to achieve proximal and distal alignment of the quadriceps and patella by muscle-tendon transposition in this patient with open physis (Fig. 7).

Reduction was maintained post-operatively for 8 weeks with a spica cast. After removal of the cast, the patient underwent a rehabilitation period.

At 15 months' follow-up, the knee is satisfactorily balanced, with no clunking and no residual patellofemoral pain.

References