SYNDROMES

Down syndrome

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Introduction

Dr. John Langdon Down, the English physician, first described this syndrome in 1866 in his article that some might consider it today as 'politically incorrect'.\textsuperscript{1} It is one of the most common chromosomal disorders. The overall life expectancy of the sufferers has increased from 9 years in 1925 to over 40 years today. As a result the orthopaedic problems are now more commonly seen.

Orthopaedic manifestations are mainly attributed to ligament laxity and include atlantoaxial instability (AAI), scoliosis, congenital hip dysplasia, hip dislocation (and secondary osteoarthritis), patellofemoral instability, pes planus, and metatarsus primus varus.\textsuperscript{2–4}

Epidemiology and genetics

The incidence of Down syndrome is approximately 1 in 700 live births increasing with maternal age. Trisomy of chromosome 21 is the most common genetic abnormality; however, cases of translocation and mosaicism were also reported.

Prenatal diagnosis and screening

High free $\beta$ chain of human chorionic gonadotrophin ($\beta$-HCG) and low pregnancy associated protein A (APAPP-A) measured in maternal blood between 8th and 14th week of gestation indicates a probable Down syndrome affected fetus. These blood tests can be combined with nuchal translucency measurement (NTM) to increase the accuracy of the diagnosis. NTM is ultrasound scanning (USS) detection of a thickened and oedematous flap of skin at the base of the neck. A measurement of more than 3 mm is considered to be abnormal (Fig. 1). NTM can detect up to 80% of Down syndrome affected fetuses during the first trimester of pregnancy.

Orthopaedics manifestations

Atlantoaxial instability

Approximately 15% of individuals with Down syndrome have AAI. However, most of these patients are asymptomatic, and only 1–2% have a serious neck problem that requires surgical intervention.

Symptomatic AAI is either due to subluxation (which can be severe enough to damage the spinal cord) or dislocation. The manifestations of symptomatic AAI include fatiguability, difficulties in walking, abnormal gait, neck pain, limited neck mobility, torticollis, incoordination, sensory deficit, and hypereflexia. Signs and symptoms often remain stable for months or years, although sometimes patients can progress to paraplegia, quadriplegia, or death.

The minimum required radiological investigations include an antero-posterior, flexion and extension lateral, and odontoid view radiographs. The atlantodens interval (ADI) is the measured distance between the posterior edge of the ring of C1 and the anterior edge of the odontoid peg (Fig. 2). It is normally less than 3.5 mm in adults.

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Treatment depends on history, physical examination, and the ADI.

Recommended treatment for AAI (ADI = atlantodens interval in mm)

| ADI 4–5  | Restriction of high-risk activities |
| ADI 6–9  | MRI or CT is recommended to investigate the presence of any neurological compromise before planning potential treatment |
| ADI ≥10  | Posterior cervical fusion and wiring |

Scoliosis

Scoliosis is common in patients with Down syndrome. Treatment is the same as in other individuals, with bracing being the initial therapy. If necessary it is followed by surgical intervention i.e. arthrodesis of the spine.5,6

Hip abnormalities

A total of 5–8% of children with Down syndrome will develop at least one form of the following hip pathologies:

- dislocation,
- developmental dysplasia (Fig. 3),
- slipped upper femoral epiphysis,
- Legg-Calve-Perthes disease,
- avascular necrosis.

Dislocation of the hip is the most common hip abnormality, occurring in 4.5% of patients (Fig. 3). Down syndrome patients have increased external rotation of the hip. The acetabulum is deep, with a horizontal roof and reduced anteversion. The femoral head sits more deeply. Mechanically the hip joint is stable; however, due to ligament laxity and increased range of movement (especially in external rotation) the hip can dislocate. The proximal femur has a normal neck-shaft angle and a moderate increased anteversion.7 Interestingly, hip subluxation in children with Down syndrome is hardly ever found at birth but instead is most common between the ages of 3 and 13 years. The most common sign of dislocation is a limp, with or without pain. Such patients also have a delay in walking and abnormal mobility of the hip joint.

The treatment for first-time dislocation begins with hip immobilisation in a cast. However, this is usually unsuccessful and frequently needs to be followed by surgical intervention. Surgical options include capsular plication, varus derotation osteotomy (as the proximal femur typically is anteverted and in valgus position) (Fig. 4), and periacetabular osteotomy.8–11

Slipped upper femoral epiphysis is more common in Down syndrome patients. This condition is often associated with obesity and hypothyroidism, both of which are common in teenagers with Down syndrome. Clinical presentation and treatment are the same as other individuals.

Legg-Calve-Perthes disease is another disorder more commonly seen in children with Down syndrome. Usually it presents with a painless limp and a decrease in range of movement. Diagnosis is made radiologically. Treatment is the same as for other patients.

Patellofemoral instability

Instability of the patellofemoral joint is seen in almost 20% of the patients with Down syndrome, but is rarely disabling. It is commonly missed and overlooked during clinical examination (Fig. 5).

Dugdale and Renshaw classification of patellofemoral instability in Down syndrome12

- Grade 1: stable patellofemoral joint
- Grade 2: patella can be subluxated laterally to more than half of the patellar width
- Grade 3: dislocatable
- Grade 4: dislocated but reducible
- Grade 5: dislocated irreducible

Fortunately, the majority of the patients are in grades 1 or 2. Patients with grades 4 or 5 are usually pain free and surprisingly patients with grade 5 PFI and good functional state were reported in the past.
Treatment depends on the function, mobility, other deformities around the knee and the grading of instability. Surgical intervention without addressing other deformities will result in osteoarthritis of patellofemoral joint. Patients with grade 3 PFI are usually the candidates for realignment surgery.

Foot abnormalities

Pes planus is seen in the vast majority of these patients. In mild cases, the heel is in a neutral position whereas in severe cases, the heel goes to valgus with the pronation and collapse of the midfoot. Flatfoot results in heavy calluses and bone spurs. Some cases respond to orthotics, but the majority are resistant to non-operative treatment and as a result surgical correction is preferred.

Metatarsus primus varus is also commonly seen in people with Down syndrome. Mild or early cases may be treated with orthotics, but severe cases require surgical correction. Surgical techniques for correction of pes planus and hallux valgus are the same as for other patients.

References