Original

Motor development in children with Down syndrome and associated osteoarticular pathology

Inmaculada Riquelme Agulló[1], Beatriz Manzanal González[2]
[1]Escuela de Enfermería y Fisioterapia. Universitat de les Illes Balears.
[2]Centro de Desarrollo Infantil y Atención Precoz del Maresme.

Correspondence:
Inmaculada Riquelme Agulló.
Escuela de Enfermería y Fisioterapia.
Universitat de les Illes Balears.
Edificio Beatriz de Pinós.
Ctra. Valldemossa km 7,5.
07122 Palma de Mallorca (Spain).

Article received: 12.05.06

Summary

In the population of children with Down Syndrome (DS), some motor patterns used more frequently can be found. The characteristics of the child with DS, such as hypotonia, joint instability, shortness of the limbs related to the length of the trunk and his neuropsychological alterations, have made him repeat motor patterns which are functional but not the most adequate. Despite the fact that these patterns are not limited exclusively to the children with DS and may be found in the normal development of other children, its repetition can sometimes produce alterations in the muscle-skeletal system. Precocious Physiotherapy helps the prevention and treatment of the possible muscle-skeletal pathology and stimulates a proper psychomotor development of the child with DS.

Key words: Development. Down syndrome. Hypotonia. Joint instability. Psychomotor disorders.

Motor development in children with Down syndrome and associated osteoarticular pathology.

1. Introduction

Motor development during the first year of life activates and perfects all of the neuromotor processes that are necessary for future adult mobility. Because of the characteristics of children with Down syndrome (DS), particularly hypotonia and joint instability, motor development will occur noticeably later than it does in normal children (for example, children with DS do not usually begin to walk before the age of two). In addition, these same characteristics mean that children with DS adopt motor patterns that compensate for what they lack in terms of strength, joint stability and the length of their limbs. If these patterns are repeated frequently, they can in the long term entrench abnormalities in the musculoskeletal system, leading to future disorders. Increased joint instability (due to less containment by joint soft tissue) means that those joints subjected to a more continuous load (the hips, knees and feet) or a great deal of mobility (atlantoaxial joint) are more affected.

2. Specific forms of motor development in children with DS

Because of hypotonia, newborn babies with DS frequently adopt what is commonly known as the "frog-leg" position. The upper limbs are close to the body with elbows flexed and hands predominantly open, with the hips wide apart in external rotation. Osteotendinous reflexes are often reduced and primitive reflexes tend to be weak.

During the first three months of life, the main challenge for babies with Down syndrome is to gain head control. When supine, lack of midline head control means that they will be unable to focus their gaze on what is happening in this area. Furthermore, hypotonia restricts upper limb movement to the horizontal plane, so that while arms can be dragged around, they cannot be raised to the child's mouth or lifted to reach for an object hanging above them. As for the lower limbs, although infants with DS will be able

to kick their feet alternately, the hips will remain markedly separated.

When these infants are placed prone, the excessive flexion of the lower limbs under the abdomen will cause an elevation of the pelvis, shifting the babies' weight entirely onto the anterior part of the body, which makes it impossible to extend the trunk or support the body on the forearms. When trying to lean on their elbows, these babies generally place them too far apart and are unable to support themselves on them; their chests sink between their upper limbs, and when they extend their heads to look around, they recline excessively and experience great difficulty extending the trunk.

Later on, lying prone, although they still find it difficult to extend their trunks and lean on their hands, they can reach things by dragging their arms along the floor. An abnormal position of the lower limbs can still be seen, with widely separated hips, flexed knees and the soles of the feet touching each other.

During the second trimester of life, infants also start to be able to sit up with the aid of a high back support, which can be lowered as they develop control and balance of the head and trunk. Obviously, they will lose their balance when attempting to reach for and handle objects.

During this period, infants will also begin to roll over; this is done very quickly in order to take advantage of momentum. Rolling usually first develops prone-to-supine as sudden head movements cause the infant to fall and roll over; however, this does not occur if the infant is resting prone on elbows set very far apart, since such support blocks the momentum. Rolling over supine-to-prone can be done in two different ways: by dropping vertically held lower limbs when the infant turns its head to look at an object, or by arching the head and trunk in extension, which propels the infant to a lateral decubitus position. The difference between these two forms of rolling over is that keeping the lower limbs vertical entails the use of the abdominal muscles, which is not necessarily the case in the second form (therefore considered an unadvisable pattern of movement) (Figure 1).

From the third trimester onwards, the infant's ability to move increases notably, which is why it is necessary to look out for the appearance of incorrect patterns of movement that may have an adverse effect on future posture and movements (for example, standing with legs splayed apart and external hip rotation, with stiff extended knees).

During this trimester, sitting up unaided is hampered by the shortness of the upper limbs: these children have to drop much further before their hands can rest on the ground, and such support creates positions with the trunk bent down very low. Progression to the sitting position does not tend to occur in the same way as in children without DS, starting from an oblique sitting position (sitting obliquely enables children to experiment with posture and movement in a way that helps them learn to lateralise body weight properly, twist the trunk and use the upper limbs to change position). Children with DS, because of their hypotonia and joint instability, sit up by propping up the trunk with the upper limbs from a starting prone position in which the hips are splayed wide open and the knees rigid and in extension. This splayed position of the lower limbs is a pattern that often persists when standing and walking (Figure 2). Getting out of the sitting position is also very complicated for infants with DS because of their instability; they prefer forward and backward movements to lateral movements, and will get out of the sitting position either by letting themselves fall backwards or by using the same incorrect pattern that they used to get into the position, flexing the trunk and separating the hips.

Children with DS experience difficulties creeping because their upper limbs are not strong enough to pull their bodies along; this means that they may ignore this form of movement and try to move by rolling over or pivoting on the abdomen. Four different patterns of creeping can be established: stretching out one arm to reach forward and propping themselves up on it to free the other arm; dragging themselves along using both upper limbs at the same time; dragging themselves along using both upper limbs alternately;

Figure 1. Starting to roll over with the lower limbs extended.

Figure 2. Sitting position with lateral rotation and extreme abduction of the hips.

or moving onto hands and knees and then dropping forwards.

Progression to quadrupedal crawling is also difficult because of the weakness of the upper limbs and the abdominal muscles. When this milestone is achieved, the weight-bearing position of the lower limbs should be carefully noted, as these tend to be too far apart with hips rotated, and the child may even be unable to lift the legs away from the abdomen in the initial stages, maintaining what is known in Spain as the "rabbit" position (Figures 3 and 4).

Normally developing children begin to kneel and use the "jockey" position, and progress to standing by pulling themselves up with the upper limbs. Children with DS have little upper-limb strength; therefore, these activities are delayed until the upper limbs are strengthened through other positions and movements. What they do find easy, however, is the so-called "bear" position (on hands and feet), as it enables them to take advantage of the tendency to separate the lower limbs (providing a wider support base) and to stiffen their knees in extension, which stabilises them. From this position, they can progress easily to the standing position by extending the trunk (Figure 5).

The upright stance tends to bring together all of the postural vices from previous stages: hips separated and externally rotated, knees rigid and in extension,

Figure 5. Moving to the standing position from the "bear" position.

Figure 3. "Rabbit" position.

Figure 4. "Rabbit" position, with hips abducted and laterally rotated.

Figure 6. Posture with anteversion of the pelvis and abdominal support.

anteversion of the pelvis and frequent propping of the abdomen because of abdominal hypotonia, which does not facilitate good trunk balance. While this posture is easier at first and helps these toddlers find their balance, it needs improvement to enable them to walk in a correct and balanced manner. Also, movements performed while standing (for example, picking objects up off the ground) are much easier if they involve extending the knees rather than flexing them (Figure 6) (1).

The majority of these children begin to walk between 19 and 24 months, but a significant number begin after 25 months (25%) (2). Joint instability and hypotonia make the lower limbs unstable. Children will adopt a pattern of walking with the limbs

separated and external rotation of the hips, hyperextension of the knees and pronation and eversion of the feet as strategies to increase stability. This pattern is not effective and, if allowed to continue, may lead to knee and foot problems. Walking will be painful and endurance will decrease (3).

3. Osteoarticular disorders

The musculoskeletal characteristics (hypotonia and joint instability) and motor control issues (coordination and balance) of people with DS cause changes to joint biomechanics in the long term. Osteoarticular deformities may sometimes be congenital but, in the majority of cases, they develop during growth. If such deformities are not prevented, adult life will be threatened by conditions such as premature osteoarthritis and considerable pain, affecting the feet in particular (4).

The joints most at risk in this population of patients are the atlanto-occipital joint, the atlantoaxial joint and the cervical spine, the lumbosacral joint, the head of the femur, the patella, and the joints of the bones in the feet, particularly the subtarsal joint (5).

3.1 Atlantoaxial instability

Atlantoaxial instability is diagnosed with lateral x-rays of the cervical spine in flexion, extension, and the neutral position, and is confirmed by the existence of a gap of 5mm or more between the atlas and the dens of the axis. Congenital laxity of the transverse ligament is believed to be the cause of the subluxation.

Its diagnosis is controversial, since the majority of those affected never experience any symptoms (asymptomatic subluxation). The most logical response is to watch out for any symptom of spinal cord compression (torticollis, changes in walking, sensory loss, muscle weakness, pyramidal signs, changes in defecation and urination, etc.). Approximately 15% of people with asymptomatic atlantoaxial subluxation identified by x-ray eventually develop symptoms (6). Quite frequently, dislocation is accompanied by congenital structural abnormalities in the occipitoatlantoaxial region (7).

People with atlantoaxial subluxation must take special precautions; contact sports, acrobatics, jumping exercises and diving are to be avoided.

Though uncommon, atlanto-occipital instability – almost always secondary to trauma – should not be overlooked (4).

3.2 Spinal deviations

Findings of scoliosis tend to be secondary to bad posture; they occur primarily in the thoracolumbar region, and are usually moderate (Figure 7).

3.3 Hip

Hypermobility and joint instability is found in the hips of children with DS because of soft-tissue laxity and hypotonia, although the degree of functional involvement varies greatly. The acetabulum may be normal or dysplastic, and x-rays typically tend to show lower-than-normal acetabular angles (8).

Despite these characteristics, the hips of children with DS show relatively little dislocation and subluxation because of the width of the iliac crests and the fact that they have an acetabular roof that is more horizontal than normal (4). In fact, congenital hip dislocation does not appear to occur more than average. However, in adolescents and adults, there does seem to be an increase in the frequency of hip dislocation, due, in addition to joint instability, to the presence of coxa valga associated with femoral anteversion. Recurrent dislocation may affect the growth of the head of the femur, causing, on the one hand, premature deterioration or the appearance of Perthes' disease and, on the other hand, femoral neck valgus deformity, which will increase with weight bearing and progressively subluxate the joint to the extent that it may cause chronic dislocation that will be difficult to correct (4).

The natural history of this hip instability tends to consist of the following stages (8):

Initial or pre-walking stage (from birth up to

Figure 7. Thoracolumbar scoliosis (30º Cobb angle).

approximately two years). In this stage, examination is clinical, revealing no abnormal signs, and hip hypermobility is frequently observed. X-rays do not display any bone abnormalities, except for some typical features such as lower-than-normal acetabular angles.

Habitual hip dislocation stage (from two years onwards). This coincides with the onset of walking. Repeated dislocation with specific movements is typical, and is just as easily reduced. Sometimes, the hips creak and pop, although X-rays reveal no abnormalities.

Limping stage. The child walks with a limp (i.e. incorrectly), and walking may be painful. X-rays demonstrate progressive acetabular dysplasia, a developmental abnormality that causes changes in the bone tissue and tends to give rise to pathological fractures and femoral subluxation.

Fixed dislocation stage. The hip is permanently dislocated, with an elevation of the femur that creates a neoacetabulum.

3.4 Knee

3.4.1 Dislocation of the patella

Recurrent patellar dislocation tends to be secondary to joint instability, associated with muscle hypotonia, which causes genu valgum. Consequently, the femorotibial axis shifts and, over time, makes the movements of the quadriceps and the patellar tendon abnormal, leading to lateral displacement of the patella (5).

Problems present in the form of patellar displacement, inflammation, inability to walk and frequent falls with secondary injuries. All of this makes postural control and overall dynamic coordination very difficult (5). Permanent dislocation can potentially lead to flexion deformities and cause the quadriceps to lose its extensor role and become an abductor and flexor, consequently giving rise to tibial dislocation (4).

This patellar hypermobility can trigger joint cartilage injuries in the form of chondromalacia patellae, which will cause pain when walking and joint effusion (4).

3.4.2 Genu valgum

Genu valgum is a frequent finding. It causes gait disorders and tends to be associated with pes planovalgus. In an upright stance, the plantar deformity shifts changes the bearing points so that the weight-bearing axes are malpositioned and the knees tend to move closer together, thus increasing genu valgum. To keep the knees from rubbing together, children with genu valgum walk either keeping their legs far apart, markedly flexing at the knee or hip, or using the scissor gait. They often lose their balance and fall as a result (Figure 8) (9).

Figure 8. Genu valgum.

3.4.3 Genu recurvatum

Muscle hypotonia and hyperlaxity also tend to cause stance and gait patterns with a marked presence of genu recurvatum. This is generally accompanied by genu valgum, lateral tibial rotation and pronated flat foot on weight bearing (9).

3.5 Foot

The feet of people with DS show quite consistent characteristics, generally pes planovalgus, which may or may not cause pain (Figure 9), and which occurs in 86% of cases.

The calcaneonavicular and interosseous talocalcaneal ligaments and the plantar muscles fail to contain the bony structures properly and alter the biomechanical structure. The talus turns upwards, forwards and medially, whilst the calcaneus is twisted into a valgus position, causing the plantar arch to collapse (2).

This results in an abnormal alignment of the lower limbs which, coupled with the structural characteristics of the other joints (such as genu valgum, genu recurvatum, etc.), greatly affects balance and, therefore, walking.

Another frequent observation is the medial deviation of the first metatarsal (*metatarsus primus*

varus). This may be accompanied by *hallux varus* (medial deviation of the first toe with subluxation of the metatarsophalangeal joint of this toe) and, later on, beginning in adolescence, *hallux valgus* (lateral deviation of the first, or great, toe) (2).

In some cases, dislocation or subluxation of the peroneal tendons has been observed; the tendons emerge from the retromalleolar sulcus and shift onto the malleolus during eversion. This condition, which is secondary to a distended peroneal tendon sheath, occurs between 25 months and five years of age, and does not create any difficulties when walking (4).

All of the above leads to significant problems when it comes to activities involving balance and coordination.

However, orthotics should not be too hastily prescribed for plantar support; these can be used once children have acquired automatic, relatively independent walking (2). In addition, it should be noted that only active exercise can keep the ankle and foot stable enough for correct gait, regardless of the appearance of the foot. The plantar muscles should be actively strengthened (5).

4. Physiotherapy and its role

The role of physiotherapy in work with children with DS is basically preventative. Rather than accelerating the speed of development, physiotherapy aims to limit insofar as possible the development of compensatory movement patterns that children with DS are inclined to resort to, and provide an opportunity to practise movements in an appropriate manner.

Children try to compensate for hypotonia, joint instability, lesser strength and shortness of limbs by developing patterns that may result in orthopaedic and functional problems (3, 10). To prevent these patterns, it is essential to start to teach posture and movements from an early age. Physiotherapy should therefore be included as one of the disciplines in DS primary care teams. In this area of practice, physiotherapy is guided by overall psychomotor development, emphasizing work on those disturbed motor features which, in the long term, can create deformities and restrict children's movement (Figure 10).

Figure 10. Balance work on a narrow beam in the physiotherapy room.

Another of the roles of physiotherapists is to train those people who spend the most time with the child to achieve the development objectives set forth at each stage. Among carers, families play a leading role, in terms of both education and teamwork. Professionals have to allay parental fears regarding their child's development, as motor retardation is the first that can be observed at an early age. Professionals also have to involve parents in the care programme, given that several studies have concluded that active parental involvement has a clearly positive effect on motor development because it provides a natural way of working on objectives so that learning is made easier.

Figure 9. Flat foot.

5. References

1. Winders P. Gross Motor Skills in children with Down Syndrome. Bethesda, U.S.A.: Woodbine House; 1997.
2. González JC. Patologia del peu en la Síndrome de Down. Síndrome de Down, Treballs i Resums Científics per a Professionals 1992; (18): 1-5.
3. Winders P. Objetivos y oportunidades de la fisioterapia para los niños con Síndrome de Down. Revista Síndrome de Down 2002; 19: 2-7.

4. González JC. Patología del aparato locomotor. En: Corretger JM, Serés A, Casaldàliga J, Trias K (eds). Síndrome de Down. Aspectos médicos actuales. Barcelona: Masson y Fundació Catalana Síndrome de Down; 2005. p. 146-62.
5. Escribá A. Síndrome de Down, Propuestas de intervención. Madrid: Gimno; 2002.
6. Pueschel SM, Findley TW, Furia J, Gallagher PL, Socla FH, Pezzullo JC. Atlantoaxial instability in Down syndrome: roentgenographic, neurologic and somatosensory evoked potential studies. J Pediatr 1987; 110: 515-21.
7. Garbayo A, Villas C. Luxación atlas-axis en el síndrome de Down. Rev Ortop Traum 1988; 32 IB: 213-7.
8. Garbayo A, Villas C, Berguiristain JL. Síndrome de Down y Luxación de Cadera. Rev Ortop Traum 1987; 31 IB: 349-56.
9. Aprendiendo sobre Síndrome de Down. http://www. downgranada.org/
10. Burns Y, Gunn P. El Síndrome de Down, Estimulación y actividad motora. Barcelona: Herder; 1995.