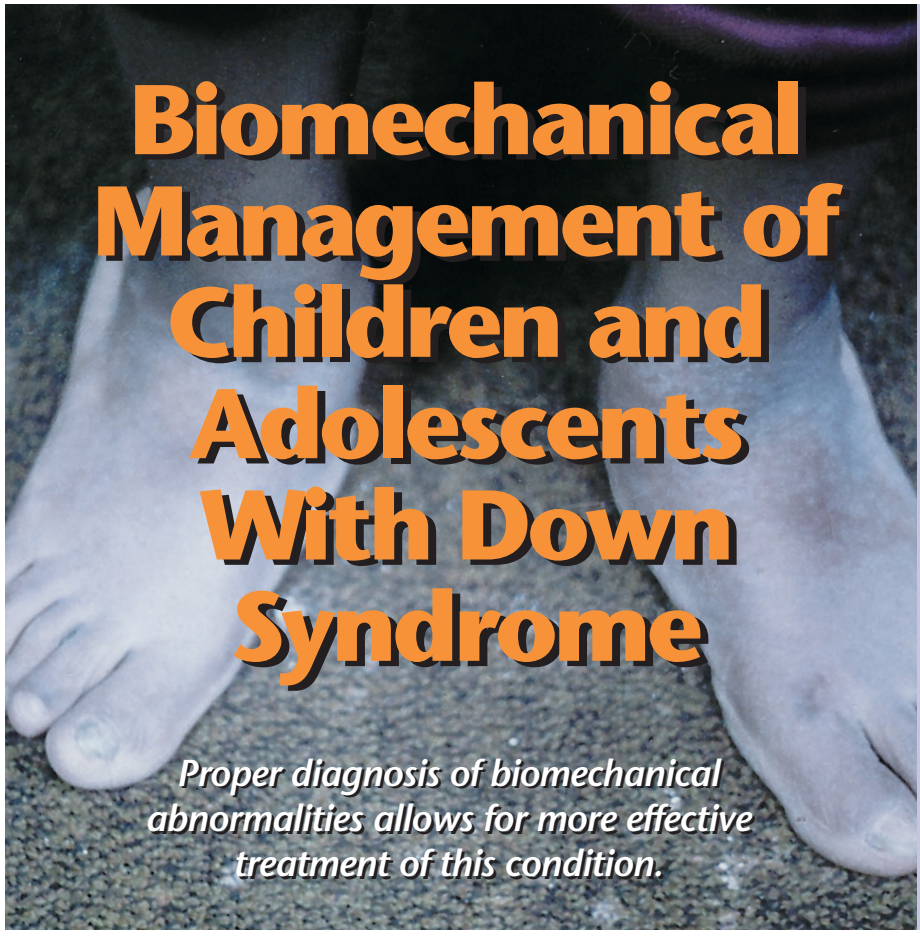
Continuing Medical Education

Biomechanical Management of Children and Adolescents With Down Syndrome



Proper diagnosis of biomechanical abnormalities allows for more effective treatment of this condition.

Objectives

After reading this article, the podiatric physician should be able to:

1) Understand the genetic abnormalities that result in Down syndrome.

2) Recognize the general characteristics of a patient with Down syndrome.

3) Take a proper medical history taking into account both common medical and orthopedic disorders that may affect ambulatory and foot function.

4) Recognize the potentially serious orthopedic/neurologic conditions associated with Down syndrome.

5) Develop a lower extremity treatment plan for a child/adolescent with Down syndrome.

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By Mark A. Caselli, DPM

Down syndrome is the most common and well-recognized congenital anomaly causing mental retardation. It occurs in all parts of the world, in all races, and in approximately one in every 700 births. It is more common in girls with a 3:1 female to male ratio.[26] Orthopedic problems are common in individuals with Down syndrome. Early recognition and treatment of these conditions can often make a significant difference in these individuals' overall ability to function and their quality of life.

The most important deformities requiring care are atlantoaxial instabil-

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Down Syndrome...

ity, dislocation of the patella, spontaneous habitual dislocation of the hip, genu valgum, and severe flexible pes planovalgus. Fifty percent of all children with Down syndrome have gait problems.[20] Painful pes planovalgus is universal in Down syndrome. Diamond et al.[8] state, "the treatment of painful feet in patients with Down syndrome is imperative, because foot pain leads to relative immobilization and immobile retarded adults do not remain long in the community". Other frequently associated medical conditions include congenital heart disease (particularly septal defect) and anomalies of the gastrointestinal tract (typically duodenal atresia and Hirschsprung's disease).

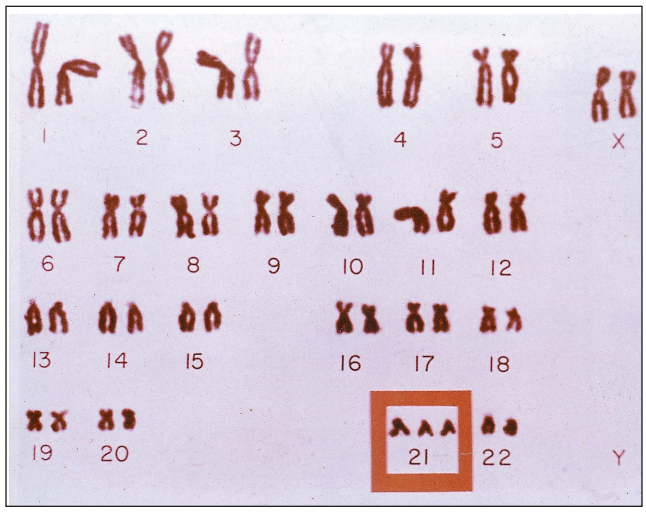

Figure 1: Karyotype of female with 21 trisomy (Down syndrome)

History

In 1866, Langdon Down identified Down syndrome in a group of mentally handicapped individuals. He was interested in the ethnologic classification of the mentally handicapped.[16,29] He thought that Down syndrome was a throwback to an ancient Mongolian ancestor. Ten years later, the connection between maternal age and Down syndrome was made. It was thought to be the result of degenerative changes of the female reproductive tract.[25,29] In 1932, Waardenburg proposed that a chromosome abnormality could explain Down syndrome.[27] In 1959, LeJeune identified Down syndrome as the first condition to be caused by an autosomal trisomy.[27]

Genetics

Most often, Down syndrome is caused by trisomy 21 in the G group. There is uneven allocation of chromosomes during normal reduction and division. Consequently, the ovum may end up with an extra G chromosome—the zygote having 47 instead of 46 chromosomes.[26] (Fig. 1)

The human genome contains approximately 100,000 genes. The number 21 chromosome contains less than 2% of the genome, or approximately 1,000 genes. The phenotype for Down syndrome is located on the 21q22 locus of the 21st chromosome. The 21q22 locus contains only about 100 genes. Therefore, only about 100 genes are responsible for the full phenotype of Down syndrome.[24]

The genes for leukemia and Alzheimer's disease are also located on chromosome number 21. This accounts for the thirty-fold increased incidence of leukemia in Down syndrome and the increased incidence of Alzheimer's disease.[22]

The cause of Down syndrome is non-disjunction of the female or male chromosome during meiotic division. Non-disjunction in the female accounts for 75% of cases and the remaining 25% is caused by male non-disjunc-

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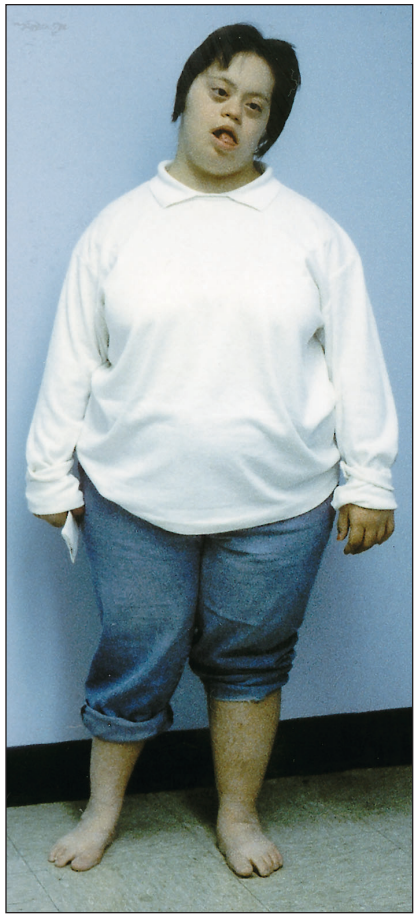

Figure 3: Characteristic facial features and stature of adolescent female with Down syndrome

Figure 2: Simian palmar crease seen in Down syndrome

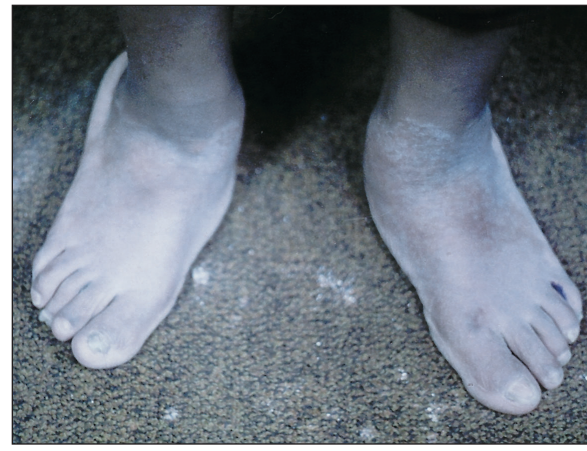

Figure 4: Severe rigid pes planovalgus in a young adult

tion. The non-disjunction can occur during any of the two phases of meiosis. The female oocytes remain in early prophase I of meiosis from birth until ovulation. The oldest eggs may be suspended in early prophase for over 40 years. These older eggs are thought to be especially vulnerable to non-disjunction. This accounts for the relationship between Down syndrome and maternal age.[3,23]

One in 2,500 babies is born with Down syndrome to women under the age of 30. One in 1,200 babies is born with Down syndrome to women between the ages of 30 and 34. The number jumps to one in 200 babies with Down syndrome born to women between the ages of 35 and 39. This is why it is recommended that pregnant women aged 35 or older undergo amniocentesis.[29]

In addition to maternal age, other causes of nondisjunction include viruses, cumulative radiation damage, history of the hepatitis B virus, grandparent maternal age, specific genes, and high thyroid autoantibody.[3]

There are three basic types of Down syndrome. The classic trisomy 21 type related to older maternal age accounts for 96% of all cases. The translocation type, known as translocation 14/21 or translocation D, occurs when the extra chromosome 21 is attached to chromosome 14. This type is not related to maternal age. Here, the chromosome number is still 46. Phenotypically, this type is identical to the trisomy category. The third type of Down syndrome is the mosaic type, which occurs in about 1% to 2% of all cases. In this group, half of the cell line has the normal 46 chromosome karyotype and half of the cell line has the trisomy 47 chromosome karyotype. Individuals in this group are phenotypically milder than those in the other two groups, and may even approach normal intelligence.[3]

The risk of recurrence of Down syndrome in a family after one affected child is about 1% for mothers in all age groups. For certain translocation or mosaic forms, the risk is much higher. Females with Down syndrome are fertile; males are sterile. Females with Down syndrome have a 50% chance of having children with Down syndrome. Identical twins with Down syndrome will both be affected. In cases of fraternal twins, usually one twin is not affected.[3]

The life span of children with Down syndrome has dramatically increased. In 1929, it was only 9 years. In 1989, the life expectancy was reported as 70 years.[25] This is because babies with Down syndrome used to die early from heart defects and respiratory infection. The heart defects are now successfully corrected by surgery and the infections are treated with antibiotics.

Characteristics

Down syndrome is usually apparent at birth, the first signs being the pronounced muscle hypotonia and the characteristic facies. The head is usually small, with few bony prominences, and brachycephalic.[15]

The eyes have a vertical epicanthal fold with slanted palpebral fissures. Normally, newborns have no true epicanthus, a fold of skin extending from the root of the nose to the median end of the eyebrow. The iris is speckled on the outside with what is known as Brushfield spots. Complications involving the eyes include myopia, cataracts, blepharitis, and ectropion (everted eyelids).

Fifty percent of all children with Down syndrome have gait problems.

The nose is small, with upturned nostrils and a flat nasal bridge. The ears are small and lack distinct contour. They are round or square, while the normal shape is oval. The upper part of the helix is folded. The lobes are small or absent. The ears are low-set. The mouth is small with droopy corners and a protruding tongue.

Even the hands are characteristic. They are short and broad, and the nails are hyperconvex. There is a dysplastic middle phalanx of the fifth finger with clinodactyly. The fingerprints tend to consist of a preponderance of ulnar loops, a distal axial triradius (85%), and in the footprints, a hallucial tibial arch (50%). There is a single line running across the palm in about 50% of patients with Down syndrome, known as the Simian crease.[2,11] (Fig. 2) Children and adolescents with Down syndrome tend to be short and heavy.(Fig. 3) They are profoundly retarded, with an IQ mean of about 50.[27]

Orthopedic Disorders

The most common orthopedic disorders seen in Down syndrome consist of metatarsus primus varus, with or without hallux abducto valgus; subluxating or dislocating patella; severe pes planus; atlantoaxial instability; scoliosis; slipped femoral epiphysis; genu valgum; and acetabular dysplasia, with or without subluxating hips.[8]

Hypotonia, ligamentous laxity, and hyperflexibility of the joints, present in 88% of children with Down syndrome, are probably the major causes of orthopedic problems.[1]

Pes planovalgus is the most common orthopedic problem in Down syndrome. It tends to be flexible and asymptomatic during the first two decades but becomes more rigid and painful during the third decade.[17] (Fig. 4) It often becomes disabling and the patient cannot wear shoes.[7] In addition, the patient has a characteristic space between the first and second toes and pronounced hallux abducto valgus. Clubfoot and syndactyly of the second and third digits may be present.[13]

Knee

Knee problems are common in Down syndrome. One study found 8.3% of institutionalized patients and 4% of non-institutionalized patients to have patellofemoral instability.[10] Knee deformities often are associated with previous foot problems. The knee problem interferes with ambulation, but is not associated with pain. The knee often gives way, causing frequent falls. Quadriceps strengthening exercises are used, but often surgery is required.[26] Genu valgum is a constant finding and is associated with various degrees of joint laxity.(Fig. 5) Chronic patellar dislocation may be associated with long-standing genu valgum in patients with Down syndrome and is not evident until the third or fourth decade.[10,17]

Hip

The hip in Down syndrome is retroverted, with excessive external ro-

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tation both in flexion and extension, resulting in out-toe gait.[13] Five percent of children with Down syndrome develop a dislocatable or dislocated hip.[1] These children are usually delayed in walking; their hips are hypermobile but not dislocatable until at two to four years of age, the affected hip spontaneously becomes dislocated and relocated. Presenting complains are a click in the hip, an increasing limp or "giving way," and refusal to walk. With recurrent dislocation, physical activity diminishes. The dislocations are not painful. If untreated, eventually subluxation or dislocation may become fixed. The recurring dislocated hip is usually treated surgically.[26]

Atlantoaxial Instability

Atlantoaxial instability is an established entity in Down syndrome.[6,14,21] It occurs in 10% to 20% of these patients. Atlantoaxial instability in Down syndrome is caused by ligamentous laxity of the transverse ligament that holds the odontoid process close to the anterior arch of the atlas. This instability results in loose joints, where the cervical vertebrae slip forward and the spinal cord is vulnerable to compression.

The neurologic manifestations of spinal compression are fatigue in walking, gait disturbance, progressive clumsiness, incoordination, spasticity, hyperflexion, clonus, and toe-extensor reflex. Onset of neck pain, headache and torticollis are indicative of malposition of the odontoid.

Individuals with Down syndrome must have a medical examination before engaging in athletic activities that involve much movement of the head and neck. In 1984, the American Academy of Pediatrics (AAP) recommended that all children with Down syndrome who participate in high-risk sports such as gymnastics, swimming, or diving be screened with lateral radiographs in neutral, flexion, and extension before beginning training or competition. If they demonstrate an abnormal odontoid or atlantoaxial interval greater than 4.5 mm, despite a normal physical exam, the AAP stated that they should be advised to avoid stressful sports.[12]

There is usually delay between onset of symptoms and diagnosis because children with Down syndrome are retarded and do not vocalize their complaints.

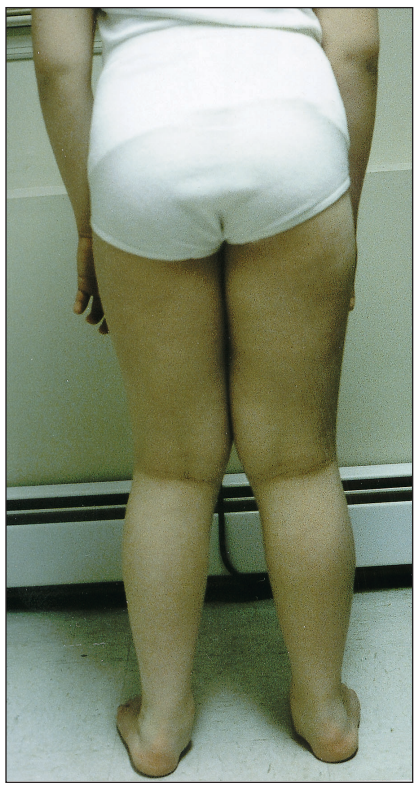

Figure 5: Adolescent patient with joint laxity, pronounced genu valgum and heel-ankle valgus

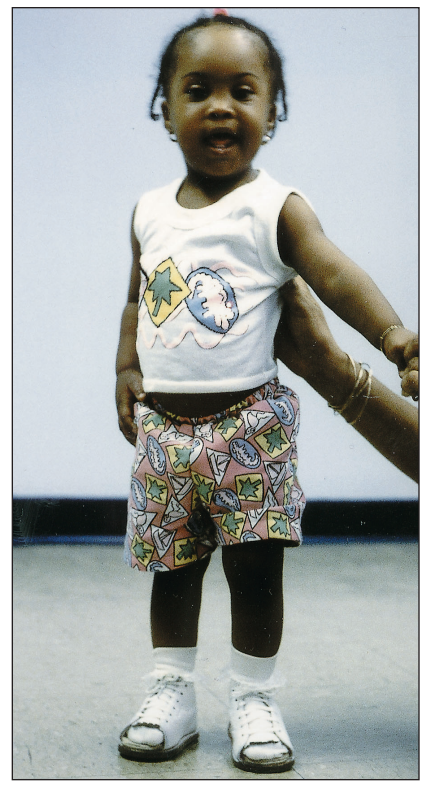

Figure 6: The open-toed straight last shoe improves foot and ankle stability for stance

Motor Development and Gait

The motor development of the infant with Down syndrome is deceptively normal for the first 6 months. However, by the age of 1 year, the infant is 4 to 5 months behind and by the age of 3 years, 10 months behind.[4] The child is late in sitting and standing.

There is a delay in independent walking between 12 and 18 months, attributable to cerebellar dysfunction with slow reaction time, hypotonia, and reduced sensory and proprioception input.[20] Children with Down syndrome, when institutionalized, begin to walk at an average age of 4.2 years, while those living at home begin walking at an average age of 2.6 years.[9]

Lindsey and Drennan[17] describe the gait of patients with Down syndrome as Chaplinesque. The hips are in external rotation, the knees are in flexion and valgus, and the tibias are externally rotated. The feet are advanced with the medial longitudinal arch as the presenting aspect of the foot and there is marked valgus and pronation of the foot.

Children with Down syndrome show a longer period of stance than independent walkers, comparable to that of the supported walking of infants. There is a decrease in hip extension and early hip extension near the end of swing. This is seen as an attempt to make a flatfoot contact instead of the initial heel contact. There is a decrease in ankle sagittal plane rotation, and exaggerated abduction of the swing limb appears to be neces-

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Figure 7: Pre-fabricated foot orthosis enhances stability of open-toed straight last shoe

sary for foot clearance.[19,20]

There also appears to be a relationship between sitting patterns and gait patterns of children with Down syndrome. Clinical observation suggests that children with Down syndrome usually have excessive external rotation and abduction of the hip, demonstrated by their sitting with widespread legs, and this excessive external rotation and abduction is displayed when they learn to sit. The wide-angled gait is caused by marked hip retroversion, genu valgum of the knee, external tibial torsion, and excessively pronated feet.[18]

It has been proven that ambulation performance, including balance and jumping, can be significantly improved in children with Down syndrome with even minimal physical therapy sessions, such as jumping classes. This type of therapy should be encouraged.[28]

Considerations in the Correction of Congenital Deformities

Early detection and treatment of congenital pedal deformities is important in a child with Down syndrome. Since these children are subject to a multiplicity of orthopedic problems, an aggressive program to maintain proper skeletal alignment can significantly decrease the severity of these problems and allow the individual to function much more efficiently. In a report on management of foot and knee deformities in the mentally retarded, Lindsey and Drennan[17] asserted that "proper alignment of the immature foot will frequently decrease the external rotation of the limb and result in development of a more appropriate gait pattern." Rather than being aggressively treated, many of the common congenital deformities, such as metatarsus adductus and tibial torsion, are overlooked because of the patients' many other medical and orthopedic problems, and no treatment is rendered.

The treatment modalities used in the correction of congenital foot and torsional abnormalities in the child with Down syndrome are the same that would be used in the normal patient. These include serial immobilization casting, corrective shoes and splints, and surgery. Due to the prolonged excessive ligamentous laxity and the relatively slower foot growth, corrective modalities often are required for longer periods of time in the child with Down syndrome. Immobilization modalities that impede walking, such as plaster casts or restrictive splinting, should be avoided in the older child, since these can further delay the progression of neuromotor development in a child that already will exhibit a significant delay in learning to walk. The use of properly modified corrective shoes should be encouraged when correcting foot pathology in children with Down syndrome who have progressed beyond the states of sitting independently and crawling.[5]

Promote Independent Stance and Walking

Although the delayed walking in children with Down syndrome is attributed to cerebellar dysfunction, much of it is also the result of excessive ligamentous laxity and hypotonia present in all of these children. This ligamentous laxity and hypotonia cause severe pronation and abduction of the feet, which greatly reduces the child's anterior-posterior stability.

The use of an open-toed straight last shoe can be beneficial in maintaining foot and ankle stability, which will promote independent stance in the child with Down syndrome. This shoe offers a rigid, flat, wide sole and a rigid heel counter, and rises to the level of the ankle, giving both foot and ankle support. (Fig. 6) The function of the shoe can be enhanced by incorporating an orthotic device to further limit pedal pronation. (Fig. 7)

There are many types of orthoses that can be used for this purpose, each with its own advantages and disadvantages. Prefabricated orthoses offer the major advantages of much lower cost to the patient and lack of necessity of taking impression casts of the patient's feet. Although they do not offer the level of pronation control that might be available with custom fabricated orthoses, there are many devices on the market that will improve the support offered by a corrective shoe alone. A prefabricated orthotic device may be desirable for the individual during the early stages of standing, when there are only short periods of weightbearing.

Custom orthotic devices offer the greatest degree of versatility in eliminating the effects of undesirable pronation. The types of foot orthoses that the author has found the most useful in the treatment of the young child with Down syndrome include leather laminates, rigid acrylic orthoses and polypropylene orthoses of the University of California, Berkeley Laboratory

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Figure 8: High top sneaker with four-ply custom molded leather orthosis. Note the deep heel seat and high medial and lateral flanges.

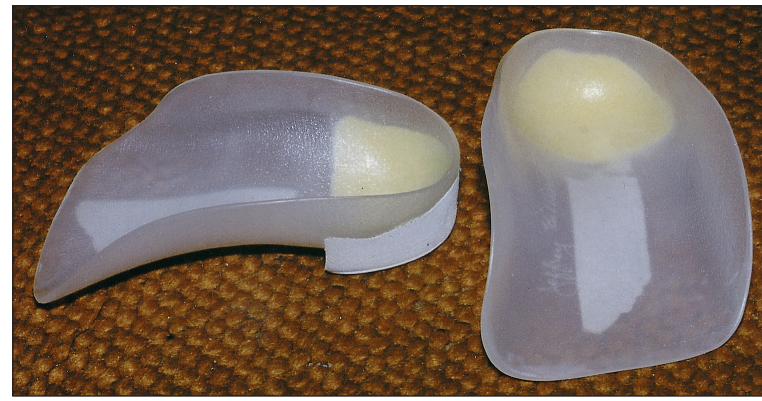

Figure 9: University of California, Berkeley Laboratory type polypropylene foot orthoses.

(UCBL) type.

Leather laminated orthoses, if used, should be ordered four-ply with a deep heel cup and high medial and lateral flanges. The leather orthoses offer the advantage of being moderately rigid with some lateral compression and expansion available to compensate for the fit of each shoe used and the ability of the orthosis-shoe combination to control the individual's pronation. When used in snug-fitting supportive shoes, the shoes will improve the function of the orthoses. If used in a more flexible or loose-fitting shoe, rather than being uncomfortable, the leather orthoses will expand laterally and offer a comfortable fit. Leather laminate orthoses also offer the advantage of being easily modified and adjusted. (Fig. 8)

Rigid acrylic devices offer excellent pronation control. They allow minimal tolerance in casting, fabrication, and shoes. Since they exhibit no lateral compression, if the device is only slightly wider than ideal, the acrylic orthoses actually can detract from the support afforded by the shoes alone.

The polypropylene orthosis (UCBL) is an excellent device, and most commonly prescribed for controlling pronation in the severe flexible flatfoot present in Down syndrome. It can be fabricated with a deep heel cup and high medial and lateral flanges. It is very thin and rigid, and yet is somewhat laterally compressible.(Fig. 9) The major disadvantage of this device is its high cost.

Supramalleolar ankle-foot orthoses can also be used. These devices require more elaborate impression-taking techniques and are expensive. The author has found that in many cases they offer little advantage over appropriate foot orthoses placed in supportive, high top, and sometimes modified shoes.

When the use of an orthotic device is contemplated in the management of the severe flatfoot deformity in Down syndrome, the practitioner must take plaster impression casts of the child's foot, being sure to accurately capture the heel and to exaggerate the correction. Foam materials used in impression taking often will lead to an improperly fitting orthosis.

Reduction of Out-Toe Gait and Genu Valgum

Many early walkers with Down syndrome exhibit clumsy, Chaplinesque gait patterns because of excessive external hip position, external tibial torsion, and severe pronation. The tripping and falling resulting from this can be reduced by improving the child's anterior-posterior stability. This can be accomplished by having the child wear high top sneakers, preferably rising above the ankle, with rigid, flat soles. Sneakers with soles that are concave from medial to lateral are most desirable. The stability of the sneaker can be enhanced further by adding a neoprene medial buttress, which effectively increases the width of its weight-bearing surface.(Fig. 10) The addition of an orthotic device will further improve the child's walking.

Pes planovalgus is the most common orthopedic problem in Down syndrome.

Severe genu valgum associated with joint laxity and severe pronation is also a common problem in Down syndrome. A goal for treatment modalities used for this problem is their ability to maintain the lower extremity in a position perpendicular to the ground. Methods of accomplishing this include the use of high top oxford shoes or, more practically, supportive sneakers, which can be modified by splitting the soling material and adding varus wedging. This modification, in combination with foot orthoses, can yield a significant improvement in lower limb alignment.

Reduction of Effects of Pedal Structural Disorders

The pedal structural problems associated with Down syndrome are the same as those associated with other patients who exhibit severe ligamentous laxity and excessive pronation. These include an increased incidence of severe hallux valgus and hammer toe deformities, as well as plantar fasciitis, fatigue, and early onset of pedal arthritis associated with severe flatfoot. These problems usually begin to manifest themselves in adolescence and early adulthood.

It is not within the scope of this article to discuss the many treatment modalities available for these problems, since they are the same as would be applied to any other patient. It is important, though, to consider that many patients with Down syndrome are aware of and concerned about shoe style, and are involved in athletic events. The author has observed that many young women with Down syndrome want to wear stylish shoes. In these cases, the consideration of a fashion-type orthosis may be appropriate. Many patients with Down syndrome are also involved in sports activities. Over 28 percent of the participants in the Special Olympics have Down syndrome. These activities must also be considered in the overall biomechanical approach to treatment. ■

Figure 10: High top sneakers with medial buttress for added stability

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Dr. Caselli is Staff Podiatrist at the VA Hudson Valley Health Care System and Adjunct Professor, Department of Orthopedic Sciences, New York College of Podiatric Medicine.

EXAMINATION

Continuing Medical Education

See instructions and answer sheet on pages 170-172.

1) Down syndrome is the most common congenital anomaly. Which of the following is the overall birth incidence of Down syndrome?
- A) One in every 50 births
- B) One in every 200 births
- C) One in every 700 births
- D) One in every 3000 births

2) Most often, Down syndrome is caused by:
- A) Trisomy 18
- B) Trisomy 21
- C) Translocation 14/21
- D) Young maternal age

3) The life-span of children with Down syndrome has dramatically increased primarily due to:
- A) Advances in cardiac surgery
- B) Early recognition of orthopedic deformities
- C) Decline in institutionalization
- D) Early identification of affected infants

4) Which one of the following is not a common finding in Down syndrome?
- A) Ligamentous laxity
- B) Spastic diplegia
- C) Hyperflexibility
- D) Hypotonia

5) The most common foot problem found in Down syndrome is:
- A) Hallux abducto valgus
- B) Hammertoes
- C) Pes cavovarus
- D) Pes planovarus

6) Patellofemoral instability is common in Down syndrome. Which one of the following is a common finding in this condition?
- A) Less common in institutionalized patients
- B) Often very painful
- C) Results in frequent falling
- D) Surgery rarely required

7) A child with Down syndrome demonstrating an increasing limp should be evaluated for:
- A) Flexible flatfoot

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EXAMINATION

(cont'd)

B) Genu valgum
C) Scoliosis
D) Dislocated hip

8) A child with Down syndrome demonstrating progressive clumsiness, spasticity, toe extensor reflex and uncoordination should be evaluated for:
A) Atlantoaxial instability
B) Severe pedal pronation
C) Peroneal spasm
D) Foot fracture

9) Motor developmental delay in Down syndrome occurs:
A) At birth
B) After 6 months of age
C) After 3 years of age
D) In early adolescence

10) The common gait pattern in Down syndrome is that of:
A) Intermittent toe-walking
B) Mild in-toe
C) Severe out-toe
D) Scissor gait pattern

11) Early treatment of foot deformities is often not initiated in children with Down syndrome since:
A) Children are mostly institutionalized
B) There are many other medical and orthopedic problems
C) The foot is not deemed important
D) The deformities are mild

12) Immobilization modalities are required to be used for longer periods of time in Down syndrome due to:
A) Ligamentous laxity and slow growth
B) Difficulty in obtaining proper fit of appliances
C) Delayed ambulation
D) Severity of deformities

13) Which one of the following causes for delayed walking in children with Down syndrome cannot be helped with orthopedic shoe gear and foot orthoses?
A) Hypotonia
B) Ligamentous laxity
C) Acetabular dysplasia
D) Severe pronation

14) Which one of the following types of orthopedic shoe gear is recommended to promote independent stance in the child with Down syndrome?
A) Sable clubfoot shoe
B) Custom molded shoe
C) High top boot
D) Open-toed straight last shoe

15) Which one of the following is not an advantage of a prefabricated foot orthosis?
A) Low cost
B) Better fit
C) No need for impression casting
D) More readily obtained

16) The advantage of using leather laminate orthoses over those made from a more rigid material is that leather:
A) Lasts longer
B) Offers more shock absorption
C) Compensates for fit in shoes
D) More durable

17) The most commonly recommended foot orthoses for the treatment of the severe flexible flatfoot seen in Down syndrome is the:
A) University of California, Berkeley Laboratory (UCBL) type
B) Leather laminate type
C) Acrylic type
D) Pre-fabricated type

18) The best method of taking an impression of a child's foot for the fabrication of orthoses is:
A) Paper tracing
B) Plaster cast
C) Foam impression
D) In shoe impression

19) Adding all but which one of the following can enhance sneaker support and stability?
A) Medial buttress
B) Varus sole wedging
C) Outer sole wedge
D) Foot orthosis

20) When treating foot pathology in a patient with Down syndrome:
A) The multiple orthopedic and medical problems must be considered
B) Because of the severe mental retardation, feedback from the patient is of little value
C) Orthopedic intervention has little effect on ambulatory outcome
D) Down syndrome is too rare to be of medical significance

SEE INSTRUCTIONS AND ANSWER SHEET ON PAGES 170-172