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Instructional lectures

Management of disabilities associated with achondroplasia

Nobuhiko Haga

Department of Pediatric Orthopedics, Shizuoka Children's Hospital, 860 Urushiyama, Shizuoka 420-8660, Japan

Abstract Achondroplasia is a skeletal dysplasia that manifests as short stature. Impairment and complications range over many disciplines including orthopedics, pediatrics, neurology, and otolaryngology. The major impairments of the extremities are short limbs, limited elbow and hip extension, and knee and leg deformities that can cause disabilities in arm function and locomotion. Hydrocephalus, a narrow foramen magnum, spinal deformity, and spinal canal stenosis can cause neurological problems, leading to disabilities in locomotion, communication, and learning. Malfunctions of the otolaryngeal system such as otitis media, upper respiratory obstruction, deafness, speech delay, and malocclusion are interrelated and can also lead to disabilities in communication and learning. Although such disabilities may cause social handicaps, most children receive a normal education. Their social maturity scale is comparable to that of normal children, but their scale of locomotion is not. The reported occupational level of female adult patients is lower than that of their unaffected siblings. When managing patients, orthopedists should consider the overall aspects of achondroplasia, including natural development and complications other than orthopedic factors.

Key words Achondroplasia · Disabilities · Complications

Introduction

Achondroplasia is a skeletal dysplasia that manifests as short stature. In the most recent international classification,[6] it belongs to the "achondroplasia group" along with thanatophoric dysplasia types I and II, hypochondroplasia, and SADDAN (severe achondroplasia, developmental delay, acanthosis nigricans). Mutations in *FGFR3* (fibroblast growth factor receptor 3) have been identified in all disorders in the "achondroplasia group." Achondroplasia is the most frequent nonlethal skeletal dysplasia, with an incidence of one per 26000 live births.[24] In Japan, 301 patients were recorded in the nationwide registration system of skeletal dysplasias of the Japanese Orthopaedic Association between 1990 and 1999. This number was second only to 345 patients with osteogenesis imperfecta.

Orthopedists usually deal with the orthopedic complications of achondroplasia. In such instances, they should consider the overall aspects of achondroplasia, including natural development and complications. The purpose of this article is to summarize the disabilities associated with achondroplasia and to help orthopedists manage them.

Disabilities associated with achondroplasia

Major or common impairment and complications associated with achondroplasia are listed in Table 1. They range over the fields of orthopedics, pediatrics, neurology, neurosurgery, otolaryngology, psychology, and rehabilitation medicine. Some of them lead to disabilities, which can be divided into those of arm function, locomotion, communication, and learning. Any of these disabilities could cause social handicaps, including those associated with education and occupation. A disablement model is shown in Fig. 1.

Management of disabilities

Short stature and orthopedic problems in extremities

Patients with achondroplasia have short-limbed short stature. The shortening clinically is rhizomelic, but radiographically the tibia/femur ratio is not significantly different from that in controls.[11] Growth charts of Japanese patients with achondroplasia can be obtained.[15,21]

Offprint requests to: N. Haga
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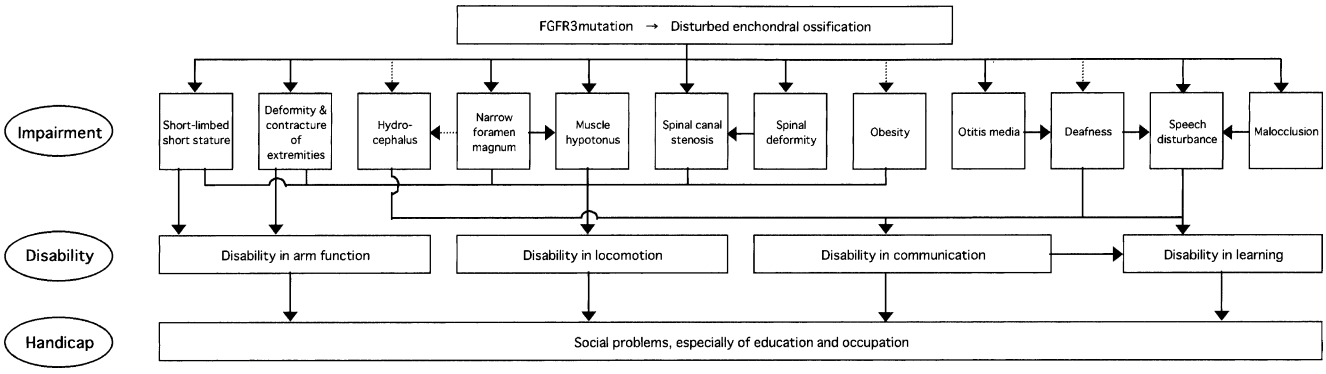

Fig. 1. Disablement model of achondroplasia. *Dotted arrows*, relation not evident

Table 1. Impairment and complications associated with achondroplasia

Extremities
Short-limbed short stature
Limitation of elbow extension
Limitation of hip extension
Knee and leg deformity
Spinal and central nervous system
Hydrocephalus
Narrow foramen magnum
Spinal deformity
Spinal canal stenosis
Otolaryngeal system
Otitis media
Upper respiratory obstruction
Deafness
Speech delay
Malocclusion
Others
Obesity

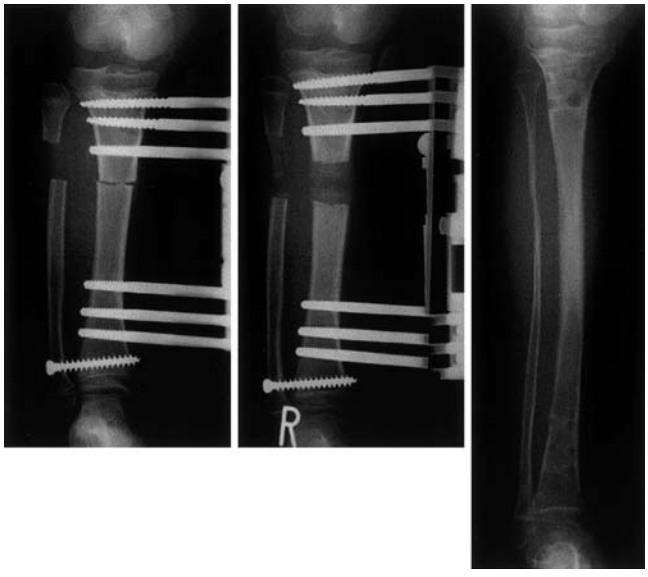

Fig. 2. Lengthening of the leg in a patient with achondroplasia

Short-limbed short stature can be treated by limb lengthening, growth hormone (GH) therapy, or both. Limbs are usually lengthened by callotasis (callus distraction) (Fig. 2).

Aldegheri and Dall'Oca[1] reported the results of bilateral femoral and tibial lengthening in 80 achondroplastic patients. The average gain in height was 20.5 cm over a treatment period of 33 months. Altogether, 31 patients experienced one or more complications, and 3 of them had sequelae of stiff ankles. Aldegheri and Dall'Oca[1] also described 10 patients who underwent humeral lengthening. The average gain was 10.2 cm over a period of 9 months. Beneficial effects of GH therapy in achondroplasia have been reported even when patients secrete normal levels of GH. An increase in height is obvious during the first 2–3 years of treatment,[22] but the long-term effects, including adult height, remain unknown.

Limitation of elbow extension is common in achondroplasia.[13] It is mainly due to a posterior convex deformity of the distal humerus. In patients with associated dislocation of the radial head the loss of extension is more severe (Fig. 3).[12] Because limitations of elbow extension rarely lead to disabilities that interfere with daily activities, treatment is unnecessary. Yasui et al.[29] surgically lengthened the humerus with simultaneous correction of a distal humeral deformity.

Limited hip extension is associated with lumbar hyperlordosis in patients with achondroplasia (Fig. 4). Neither of these problems usually leads to disabilities or requires aggressive treatment. Vilarrubias et al.[27] simultaneously lengthened the femur and corrected lumbar hyperlordosis by femoral realignment and muscle release.

Bowleg deformity is common with achondroplasia (Fig. 5).[13] Varus deformity of the knee and proximal tibia is due to proximal fibular overgrowth and associated laxity of the lateral collateral ligament, and varus

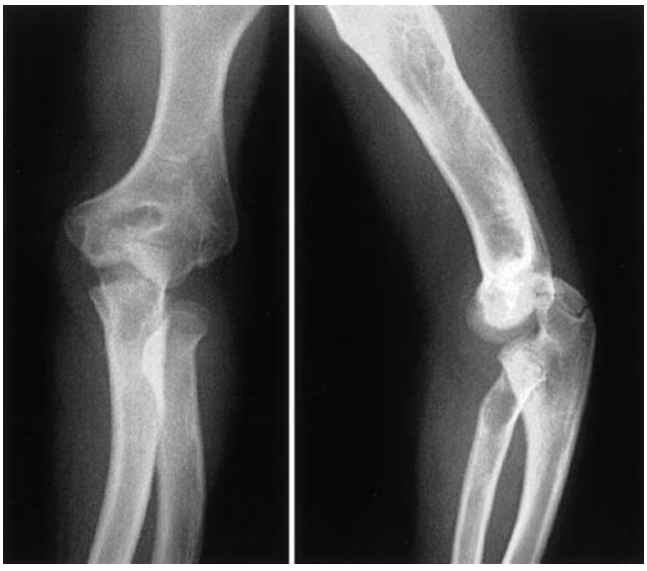

Fig. 3. Posterior convex deformity of the distal humerus with posterolateral dislocation of the radial head

deformity of the distal tibia is due to distal fibular overgrowth.[19] Osteoarthritis of the knee in adult patients has been observed only occasionally.[13] The treatment of bowleg deformity associated with achondroplasia is controversial. The effects of brace treatment and fibular epiphyseodesis have not been proved.[19] The indication of tibial osteotomy is not clear, but it has usually been done to alleviate pain and for cosmesis. About 10%–22% of patients undergo tibial osteotomies.[9,13] Varus deformity of the tibia can also be corrected during a limb-lengthening procedure.[28]

Spinal and central nervous system problems

Hydrocephalus and a narrow foramen magnum are important complications, especially during the newborn and infantile periods. Hydrocephalus is usually communicating and may relate to intracranial venous hypertension.[20] Because patients with achondroplasia have a relatively large head and frontal bossing, hydrocephalus must be diagnosed with care. Close monitoring of the head circumference and motor development compared with standard data for achondroplasia[8,15,25] leads to a justified indication for ventricular shunting. About 5% of patients undergo shunting for neurological signs.[9] Patients with increased intracranial pressure but no neurological signs have been reported.[4] A narrow foramen magnum can cause sudden infant death, sleep apnea, respiratory disorders, myelopathy, syringobulbiamyelia, and hydrocephalus (Fig. 6).[7] This may be also related to muscular hypotonia. Sleep studies must distinguish sleep apnea due to a narrow foramen from that due to other causes.[14,23] The indication of foramen magnum decompression is controversial,[26] and about 3.6% of children under 18 months of age undergo cervicomedullary decompression.[9]

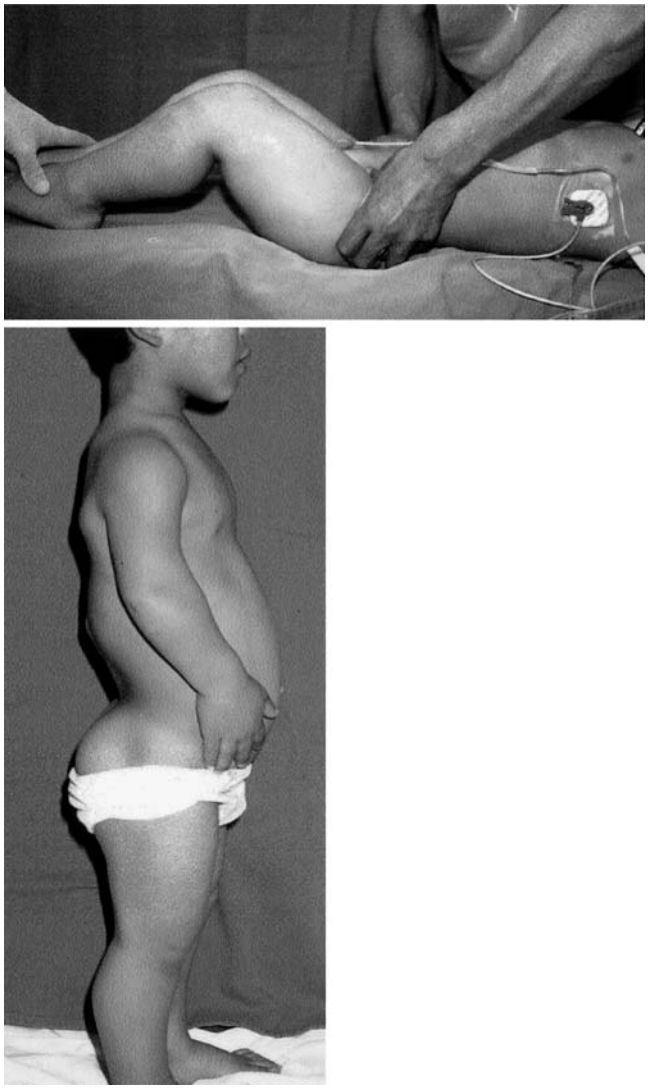

Fig. 4. Limited hip extension and lumbar hyperlordosis

Spinal deformity and spinal canal stenosis similarly cause neurological symptoms. In the sagittal plane alignment, kyphosis is usually evident at the thoracolumbar junction, as is lumbar hyperlordosis. Coronal plane alignment is usually normal, although some patients have lumbar scoliosis. As the spinal canal is narrow due to disturbed enchondral ossification, exaggerated angulation of the spine can lead to neurological deficits (Fig. 7). Preventing fixed kyphosis is extremely important in children. The kyphotic deformity usually becomes apparent while assuming a sitting posture and resolves spontaneously. However, because up to 15% of adult patients have persistent kyphosis at the thoracolumbar junction, an orthosis is usually recommended to reduce it when the deformity is marked at a sitting

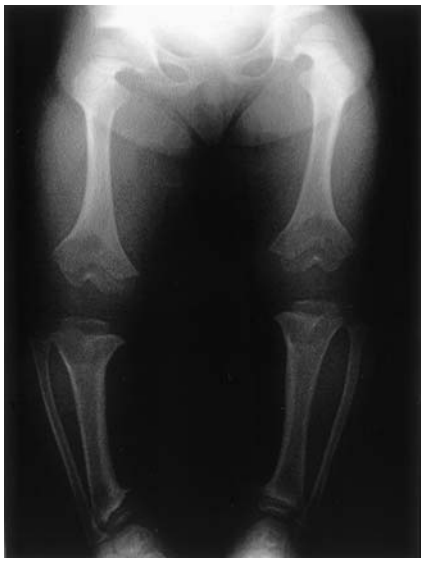

Fig. 5. Bowleg deformity in a 4-year-old patient. Note the distal fibular overgrowth in the right leg

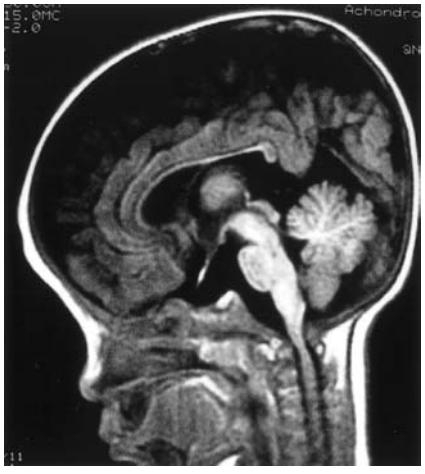

Fig. 6. Narrow foramen magnum with syringobulbia-myelia in a 4-month-old patient

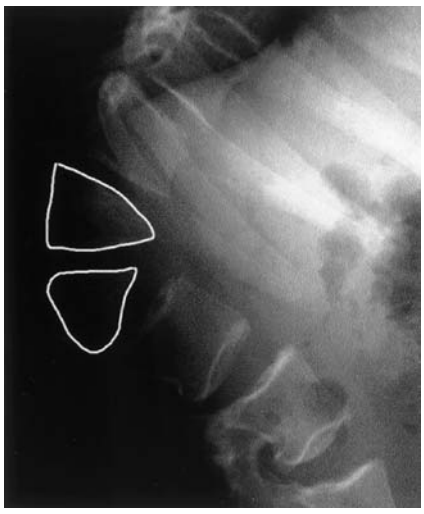

Fig. 7. Severe kyphosis at the thoracolumbar junction

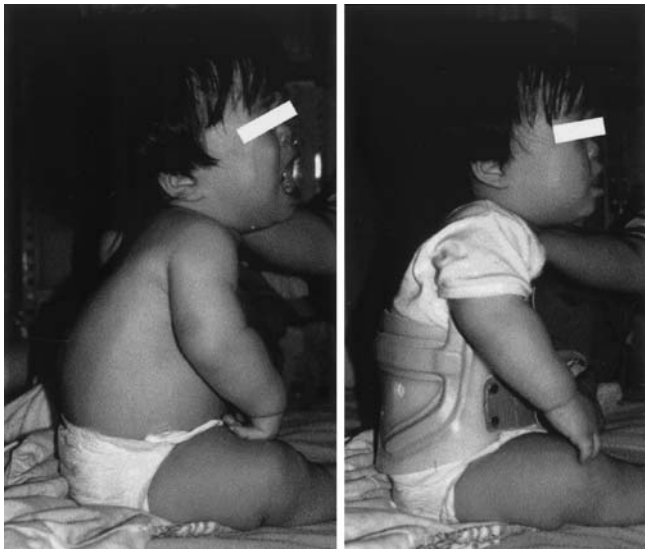

Fig. 8. Prevention of thoracolumbar kyphosis with an orthosis

posture (Fig. 8).[18] Pauli et al.[16] eliminated progressive kyphotic deformity by early prohibition of unsupported sitting; and, in those for whom such prohibition proves insufficient, he suggested a modified thoracolumbosacral orthosis.

Symptomatic spinal canal stenosis usually develops during adulthood.[9] The presence of thoracolumbar kyphosis, an L1 interpediculate distance of less than 20 mm and an L5 interpediculate distance of less than 16 mm, and severe structural lumbar lordosis are risk factors for developing disabling symptoms including spinal claudication.[10] The type of surgical procedure depends on the age of the patient, the extent of the compression, and the presence or absence of thoracolumbar kyphosis.[26]

Otolaryngological problems

Otitis media is a common complication of achondroplasia. It affects 90% of children by 2 years of age, about half of whom undergo surgical placement of a ventilation tube.[9] Relative adenotonsillar hypertrophy due to mid-facial hypoplasia causes upper respiratory obstruction, leading to obstructive sleep apnea. Otitis media and adenotonsillar hypertrophy may each cause conductive hearing loss. Speech development is usually normal,[25] but deafness and malocclusion can lead to speech disturbances. Brinkmann et al.[3] reported retarded speech development and impaired verbal comprehension. Such otolaryngological problems may lead to disabilities in communication and learning.

Social problems

Social problems are rarely associated with achondroplasia. Most children with this condition receive a normal education,[5,17] although Brinkmann et al.[3] reported lower school grades in language-related subjects. The social maturity scale of children with achondroplasia is comparable to that of normal children, but the scale of locomotion differs.[5] The occupational level of female patients was lower than that of their unaffected sisters.[17]

Conclusions

Patients with achondroplasia often consult orthopedists because of their short stature or orthopedic complications, but the impairment and complications are not restricted to care by orthopedists. This article presented an outline of health supervision for achondroplasia. The time schedule for following up patients can be obtained from the guidelines published by the American Academy of Pediatrics Committee on Genetics.[2]

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