

CLINICAL REPORT

Guidance for the Clinician in Rendering Pediatric Care

Tracy L. Trotter, MD; Judith G. Hall, OC, MD; and the Committee on Genetics

Health Supervision for Children With Achondroplasia

ABSTRACT. Achondroplasia is the most common condition associated with disproportionate short stature. Substantial information is available concerning the natural history and anticipatory health supervision needs in children with this dwarfing disorder. Most children with achondroplasia have delayed motor milestones, problems with persistent or recurrent middle-ear dysfunction, and bowing of the lower legs. Less often, infants and children may have serious health consequences related to hydrocephalus, craniocervical junction compression, upper-airway obstruction, or thoracolumbar kyphosis. Anticipatory care should be directed at identifying children who are at high risk and intervening to prevent serious sequelae. This report is designed to help the pediatrician care for children with achondroplasia and their families. *Pediatrics* 2005;116:771–783; *achondroplasia, short stature, children, health supervision.*

ABBREVIATIONS. OFC, occipital-frontal circumference; CT, computed tomography.

INTRODUCTION

This clinical report is designed to assist the pediatrician in caring for children with achondroplasia confirmed by radiographs and physical features. Although pediatricians usually first see children with achondroplasia during infancy, occasionally they are called on to advise a pregnant woman who has been informed of the prenatal diagnosis of achondroplasia or asked to examine a newborn to help establish the diagnosis. Therefore, this report offers advice for these situations as well.

Substantial new information has appeared since publication of the first policy statement on health supervision of children with achondroplasia.¹ In particular, a great deal has been learned about the molecular genetics of the disorder.² In addition, a more complete understanding of how certain serious complications can be minimized or avoided has accrued.³ The new information is incorporated into this report, which is a revision of the original policy statement.

Achondroplasia is the most common condition associated with severe disproportionate short stature.⁴ The diagnosis can usually be made on the basis of clinical characteristics and very specific features on

radiographs, which include contracted base of the skull, square shape of the pelvis with a small sacrosciotic notch, short pedicles of the vertebrae, rhizomelic (proximal) shortening of the long bones, trident hands, a normal-length trunk, proximal femoral radiolucency, and (by midchildhood) a characteristic chevron shape of the distal femoral epiphysis. Other rhizomelic dwarfing disorders such as hypochondroplasia and thanatophoric dysplasia are part of the differential diagnosis, but achondroplasia usually can be distinguished from them because the changes in hypochondroplasia are milder and the changes in thanatophoric dysplasia are much more severe and invariably lethal. Achondroplasia is an autosomal dominant disorder, but approximately 75% of cases represent new dominant mutations. Achondroplasia is caused by mutation in the gene that codes for the fibroblast growth factor receptor type 3 (*FGFR3*).^{5–7} Because virtually all of the causal mutations occur at exactly the same place within the gene,⁷ molecular testing is straightforward. It is not necessary to perform molecular testing in every child with a clinical diagnosis of achondroplasia. However, *FGFR3* testing should be performed in children who are in any way atypical or in circumstances in which differentiation from similar disorders, such as hypochondroplasia, is not certain. Such children also should be referred for clinical genetic evaluation.

A great deal is known about the natural history of achondroplasia that can be shared with the family.^{3,8} The average adult height in achondroplasia is approximately 4 ft for men and women (Figs 1 and 2).⁹ The most common complication, occurring in adulthood, is related to lumbosacral spinal stenosis with compression of the spinal cord or nerve roots.^{10,11} This complication is usually treatable by surgical decompression if it is diagnosed at an early stage.

Most children with achondroplasia do well. However, children affected with achondroplasia commonly have delayed motor milestones (Fig 3),^{12,13} otitis media, and bowing of the lower legs.¹⁴ Less commonly, infants and children may have serious health consequences related to hydrocephalus, craniocervical junction compression, upper-airway obstruction, or thoracolumbar kyphosis. Although they are less common, anticipatory care should be directed at identifying children who are at high risk and intervening to prevent serious sequelae. Most individuals with achondroplasia are of normal intelligence and are able to lead independent and pro-

The guidance in this report does not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

doi:10.1542/peds.2005-1440

PEDIATRICS (ISSN 0031 4005). Copyright © 2005 by the American Academy of Pediatrics.

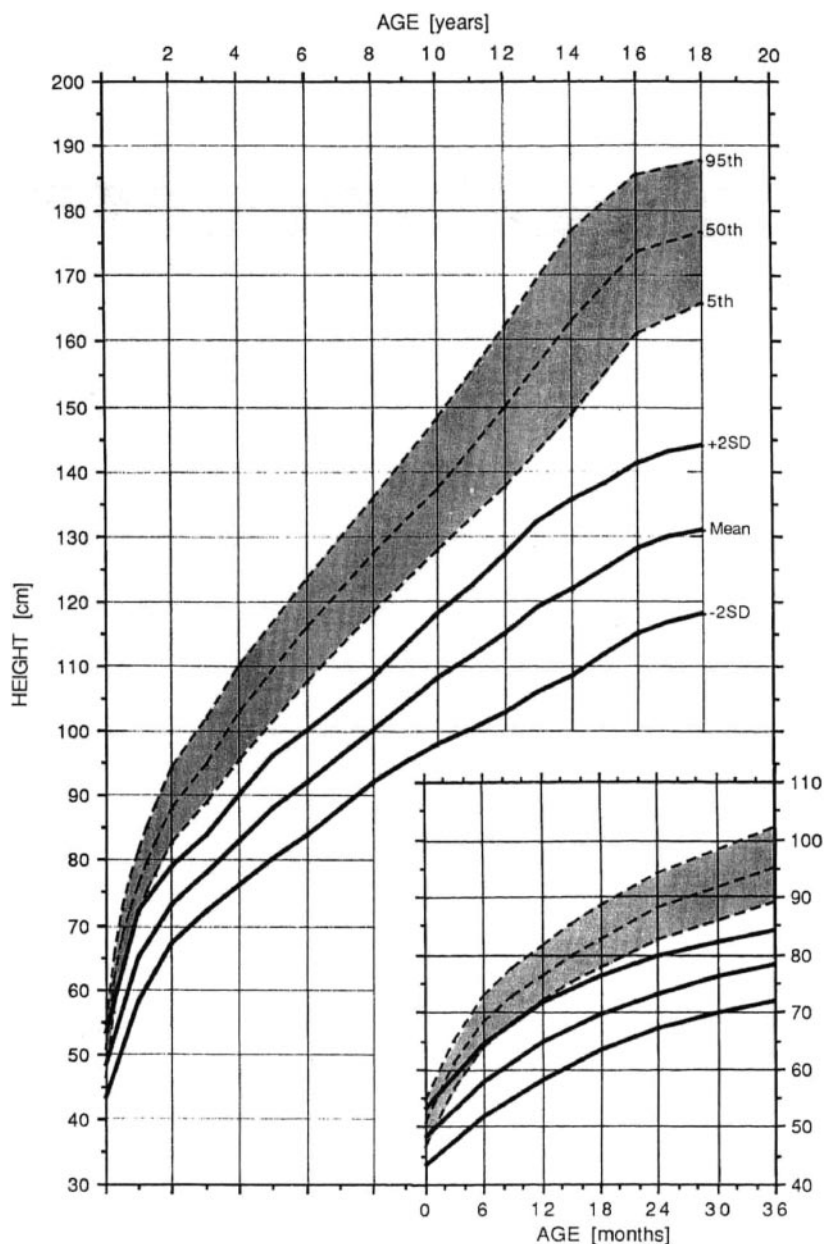


Fig 1. Height for males with achondroplasia (mean \pm 2.8 standard deviation) compared with normal standard curves. The graph was derived from 189 males. (Reproduced with permission from *J Pediatr.* 1978;93:435-438.)

ductive lives.¹⁵ Because of their disproportionate short stature, however, a number of psychosocial problems can arise. Families can benefit from anticipatory guidance and the opportunity to learn from other families with children of disproportionate short stature.

The consensus-based guidance in this report is designed to help the pediatrician care for children with achondroplasia and their families. Issues that need to be addressed at various ages are discussed (Table 1). These suggestions are not appropriate for other chondrodysplasias, because each type has its own natural history, complications, and specific guidelines. Irrespective of the availability of the guidance in this report, it is important that pediatricians and parents also consult a physician with special experience and expertise concerning achondroplasia early in the child's life, because this report only provides generally applicable suggestions that

must be tailored to a particular child's condition and needs.

THE PRENATAL VISIT

Pediatricians may be called on to counsel expectant parents whose fetus has achondroplasia or is suspected to have achondroplasia because of recognition on ultrasonography of disproportionate small stature. In some settings, the pediatrician will be the primary resource for counseling a family. At other times, counseling may already have been provided to the family by a clinical geneticist and/or the obstetrician. Because of a previous relationship with the family, however, the pediatrician may be called on to review this information and assist the family in the decision-making process.

The diagnosis of achondroplasia in the fetus is made most often with certainty when 1 or both parents have this condition. In this circumstance, the

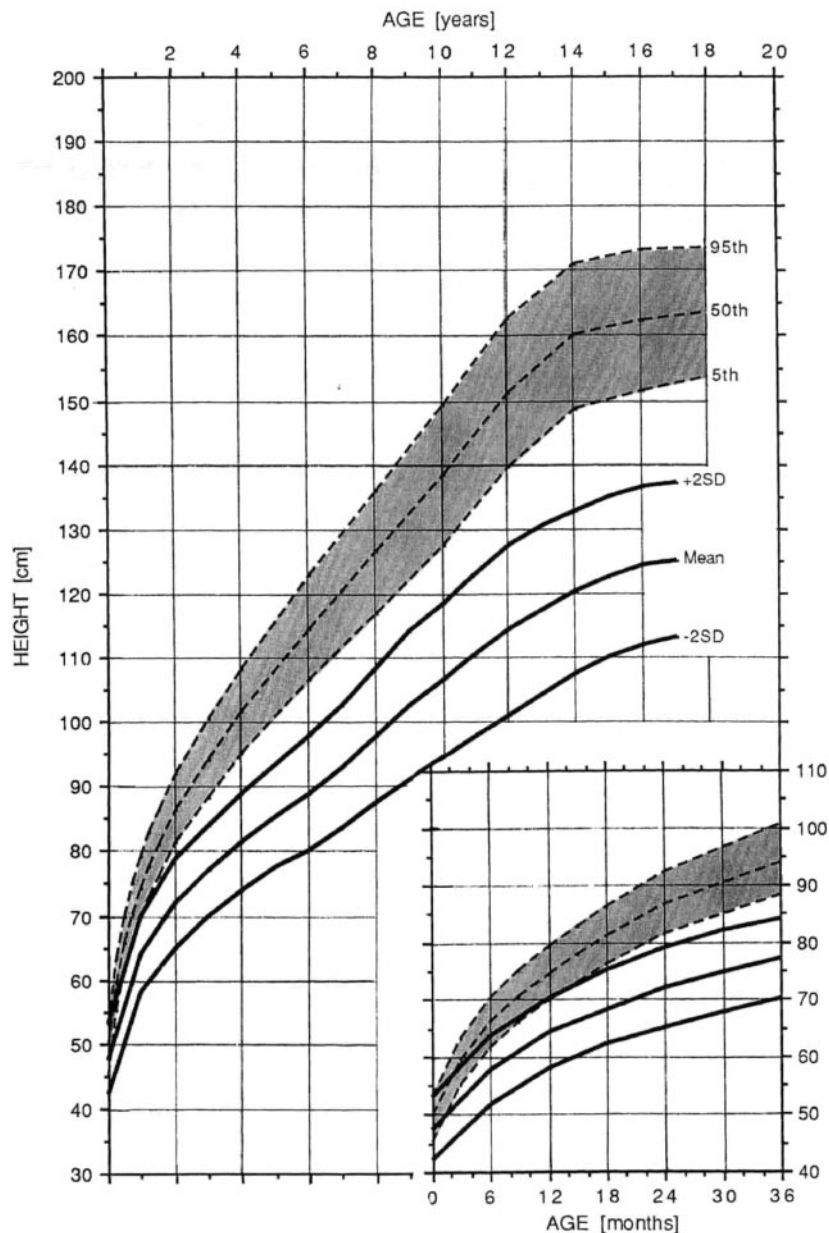


Fig 2. Height for females with achondroplasia (mean \pm 2.8 standard deviation) compared with normal standard curves. The graph was derived from 214 females. (Reproduced with permission from *J Pediatr.* 1978;93:435-438.)

parents are usually knowledgeable about the disorder, the inheritance, and the prognosis for the offspring. More often, diagnosis of achondroplasia is first suspected late in gestation on the basis of long-bone foreshortening incidentally discovered by ultrasonography. With the frequent use of ultrasonography, many cases of achondroplasia are first identified prenatally (after 26 weeks of gestational age). However, disproportionately short limbs are observed in a heterogeneous group of conditions. Misdiagnosis and inaccurate prenatal counseling of families is common.¹⁶ Confirmation of diagnosis based on ultrasonographic features characteristic of achondroplasia can be provided by molecular testing (*FGFR3* mutational testing) of prenatal specimens. If no such confirmation has yet been completed, caution should be exercised when counseling the family. In this circumstance, the pediatrician should discuss the tentative nature of the diagnosis and alternatives that may explain the identified features. The pediatrician

should also discuss the natural history of achondroplasia, because it is the most likely explanation for the findings. In cases in which the diagnosis is unequivocally established either because of the familial nature of the disorder or by prenatal molecular diagnosis (chorionic villus sampling at 11–13 weeks' gestation or amniocentesis after 15 weeks' gestation), the pediatrician may consider the following steps as appropriate.

1. Review, confirm, and demonstrate laboratory or imaging studies leading to the diagnosis.
2. Explain the mechanisms for occurrence of achondroplasia in the fetus and the recurrence risk for the family.
3. Remember that at least 75% of cases of achondroplasia occur in families in which both parents have average stature. In those cases, achondroplasia in the offspring occurs because of a mutation in the gene.

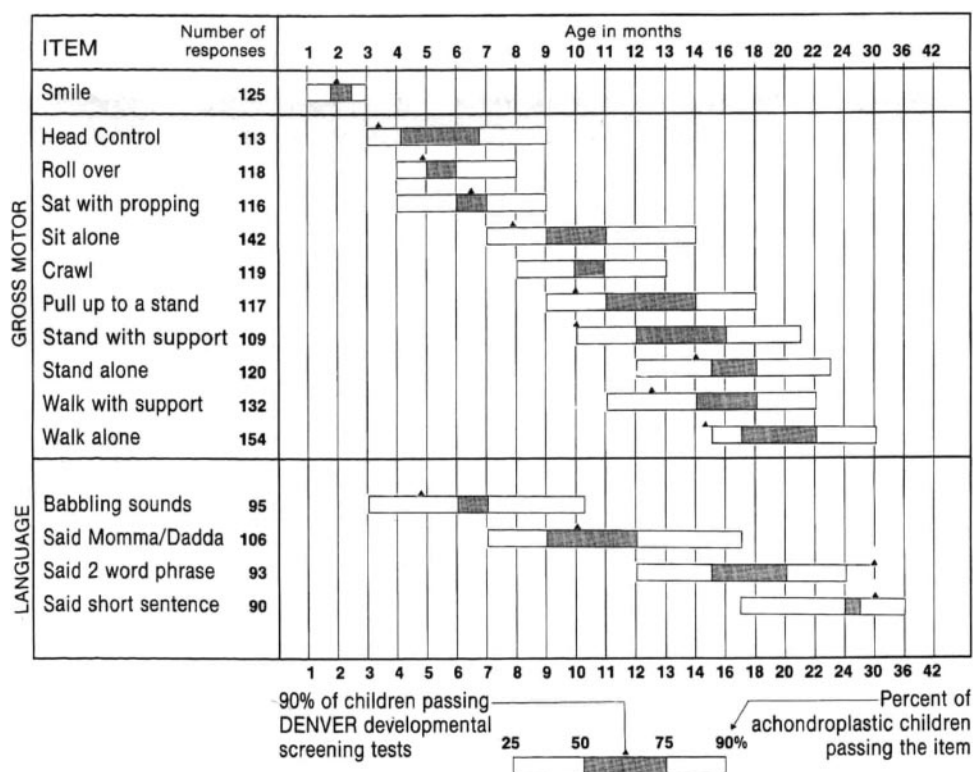


Fig 3. Developmental screening tests in achondroplasia. The bar scale shows the percentage of achondroplastic children passing the item; the black triangle on top of the bar shows the age at which 90% of normal children pass the same item. The graphs were derived from 197 affected individuals, obtained by questionnaire. (Reproduced with permission from *Am J Med Genet.* 1981;9:19-23.)

- Review the natural history and manifestations of achondroplasia, including variability.³
- Discuss additional studies that should be performed, particularly those to confirm the diagnosis in the newborn period. If miscarriage, stillbirth, or termination occurs, confirmation of diagnosis is important for counseling family members about recurrence.
- Review currently available treatments and interventions. This discussion needs to include the efficacy, complications, adverse effects, costs, and other burdens of these treatments. Discuss possible future treatments and interventions.
- Explore the options available to the family for the management and rearing of the child using a non-directive approach. In cases of early prenatal diagnosis, these options may include discussion of pregnancy termination, continuation of pregnancy and rearing of the child at home, foster care, or adoption. If adoption is planned to another family, contact may be made with the Little People of America adoption service.¹⁷
- If the mother is affected with achondroplasia, inform her that a cesarean delivery must be performed because of the characteristic small pelvis.¹⁸ A mother affected with achondroplasia may develop respiratory compromise during the third trimester of pregnancy, so baseline pulmonary function studies should be performed. Homozygous achondroplasia can be diagnosed prenatally with molecular testing of the fetus, by either chorionic villus sampling or amniocentesis. A pregnancy at risk of homozygosity should be followed

- with ultrasonographic measurements at 14, 16, 18, 22, and 32 weeks of gestation to distinguish homozygosity or heterozygosity from normal growth patterns in the fetus.
- When both parents are of disproportionate short stature, assess the possibility of double heterozygosity¹⁹ or homozygosity for achondroplasia. Some forms of double heterozygosity lead to life-threatening problems¹⁹; infants with homozygous achondroplasia usually are stillborn or die shortly after birth.²⁰

HEALTH SUPERVISION FROM BIRTH TO 1 MONTH OF AGE: NEWBORNS

Examination

- Confirm the diagnosis by radiographic studies (the diagnosis of approximately 20% of patients with achondroplasia has been delayed in the past because it was not suspected on physical examination in the newborn period, and consequently, no radiographs were obtained).
- Document measurements, including occipital-frontal circumference (OFC), body length, and body weight; plot these measurements on achondroplasia-specific growth charts (Figs 1, 2, and 4-7). Review the phenotype with the parents and discuss the specific findings with both parents whenever possible.
- The OFC should be measured at every pediatric contact during the first year (Figs 4 and 5).

TABLE 1. Achondroplasia Guidelines for Health Supervision

	Prenatal		Infancy, 1 mo to 1 y of Age				Early Childhood, 1 to 5 y of Age				Late Childhood		Adolescence
	Neonatal	2 mo	4 mo	6 mo	9 mo	12 mo	15 mo	18 mo	24 mo	3 y	4 y	5 to 13 y, Annual	13 to 21 y, Annual
Diagnosis													
Radiography													
Review phenotype													
Review proportions													
Molecular testing [FGFR3]													
Genetic counseling													
Early intervention	X												X
Recurrence risks	X												X
Reproductive options												X	X
Family support	X							X				X	X
Support groups	X							X				X	X
Long-term planning								X				X	X
Medical evaluation													
Growth/weight/OFC	X	X	X	X	X	X	X	X	X	X	X	X	X
Orthopedic consult													
Neurology consult													
Hearing													
Social readiness						X R		X R		X R		X R	
Orthodontics						S		S		R		R	
Speech													
Medical evaluation													
Radiography, only to make diagnosis or if complication													
CT/MRI brain/cervical spine	X												
Polysomnography													
Social adjustment													
Psychosocial													
Behavior and development	S/O	S/O	S/O	S/O	S/O	S/O	S/O	S/O	S/O	S/O	S/O	S/O	S/O
School													O
Sexuality													X

These guidelines ensure compliance with AAP recommendations for preventive pediatric health care. FGFR3 indicates fibroblast growth factor receptor type 3; X, to be performed; S, subjective, by history; O, objective, by a standard testing method; R, discuss referral to a specialist; →, continue to monitor.

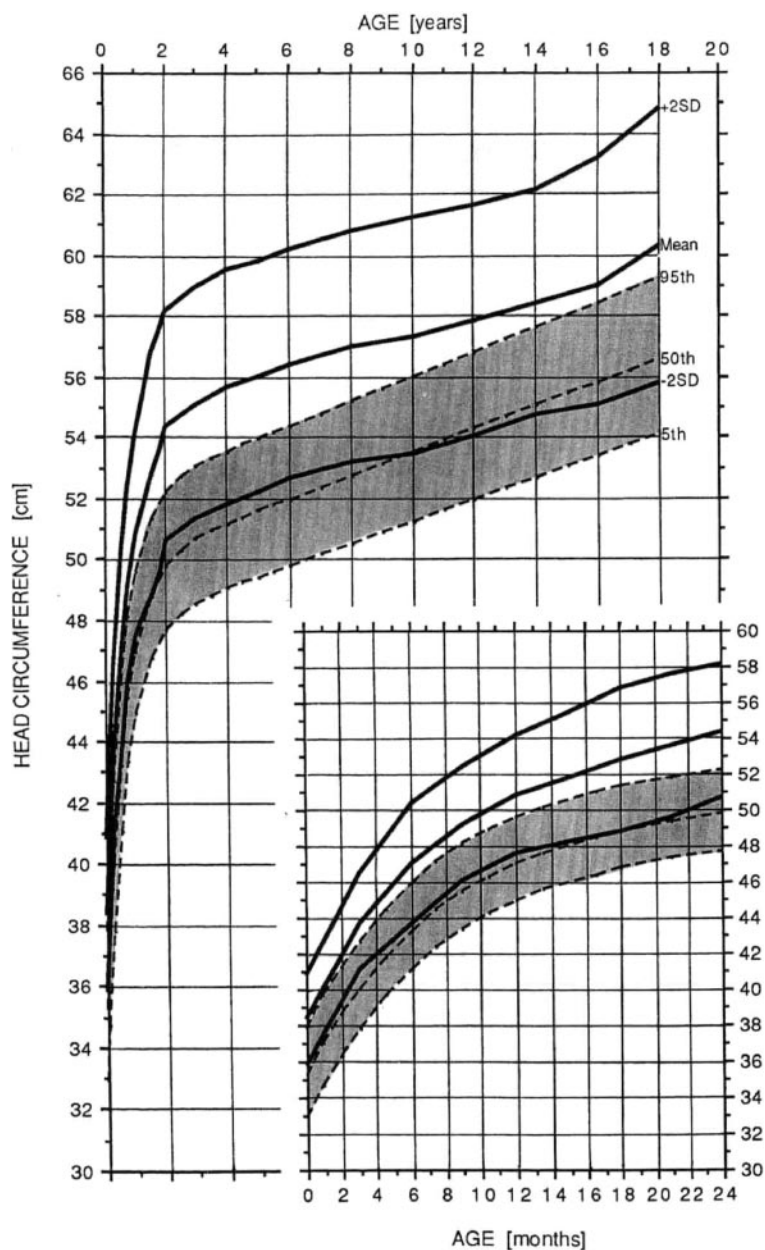


Fig 4. Head circumference for males with achondroplasia compared with normal curves (dashed lines). The graph was derived from 189 males. (Reproduced with permission from *J Pediatr.* 1978;93:435-438.)

Anticipatory Guidance

1. Discuss the specific findings of achondroplasia with the parents, including the following:
 - Autosomal dominant inheritance: approximately 75% of cases are new mutations. Germ-line mosaicism (in which some germ cells are derived from a normal cell line and some are from a cell line with a mutation) has been reported, but the risk of recurrence in sporadic cases is less than 1%.²¹⁻²³
 - Most individuals with achondroplasia have normal intelligence and normal life expectancy.
 - Although serious problems may arise during infancy, such problems affect only 5% to 10% of infants with achondroplasia.
 - Growth hormones, other drug therapies, and food or vitamin supplements are not effective in

significantly increasing stature. Growth-hormone therapy may result in a transient increase in growth rate. However, the salutary effects diminish with continued treatment. No study has clearly demonstrated a significant benefit with respect to ultimate adult stature.^{24,25} If elected, such treatment should be considered only within a research setting. Extended limb lengthening using a variety of techniques has been used far more elsewhere than in North America. It can result in substantial increases in ultimate height.^{26,27} However, it is arduous, not without risk, and costly. Most families alternatively choose to modify the environment to accommodate the child rather than the converse.

- Special achondroplasia growth curves and infant development charts have been developed (Figs 1-7); the final expected adult height for

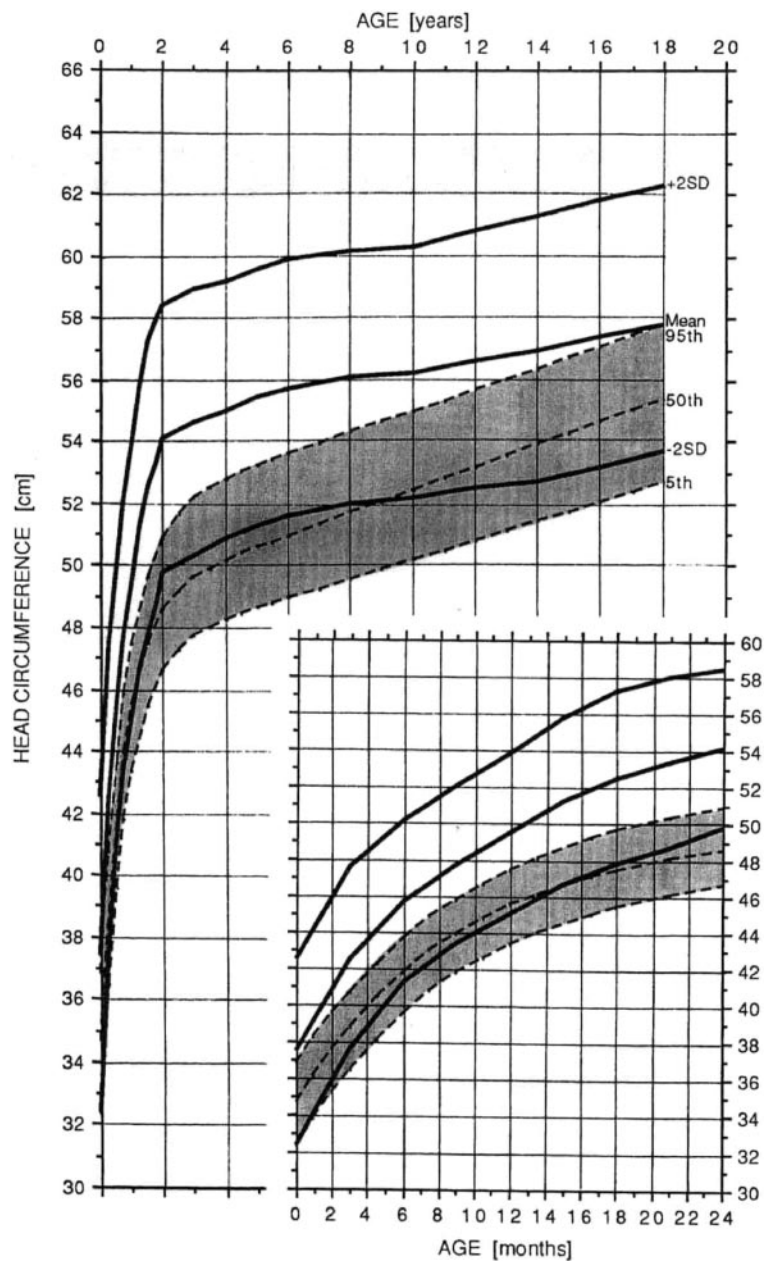


Fig 5. Head circumference for females with achondroplasia compared with normal curves (dashed lines). Data were derived from 145 females. (Reproduced with permission from *J Pediatr.* 1978;93:435-438.)

persons with achondroplasia is approximately 4 ft.⁹

2. Discuss the following possible severe medical complications and methods of prevention:
 - Unexpected infant death occurs, in the absence of aggressive evaluation, in approximately 2% to 5% of all infants with achondroplasia.^{28,29} This seems to result from central apnea arising secondary to compression of arteries at the level of the foramen magnum.²⁸ In addition, the universally small foramen magnum may result in a high cervical myelopathy.^{30,31} However, with appropriate assessment and intervention, both risks can be minimized.³¹ Parents should be advised to use an infant seat or infant carrier that has a firm back that supports the neck and to use a rear-facing car safety seat for as long as possible. They should be counseled to avoid use of products like mechanical swings and carrying

slings to limit uncontrolled head movement around the small foramen magnum. There are instances in which infants with achondroplasia who showed no clinical abnormality by examination and who were asymptomatic have died from this complication. Given this, and because it can be life saving, care of every infant with achondroplasia should include assessment for craniocervical junction risks, which includes careful neurologic history and examination, neuroimaging, and polysomnography.³¹ Neuroimaging can be by computed tomography (CT) with thin cuts and bone windows³¹ or magnetic resonance imaging (MRI),^{32,33} each of which has benefits and disadvantages: CT allows direct comparison of foramen magnum size with published achondroplasia standards and often can be accomplished without sedation or anesthesia but does not allow direct visualization of the

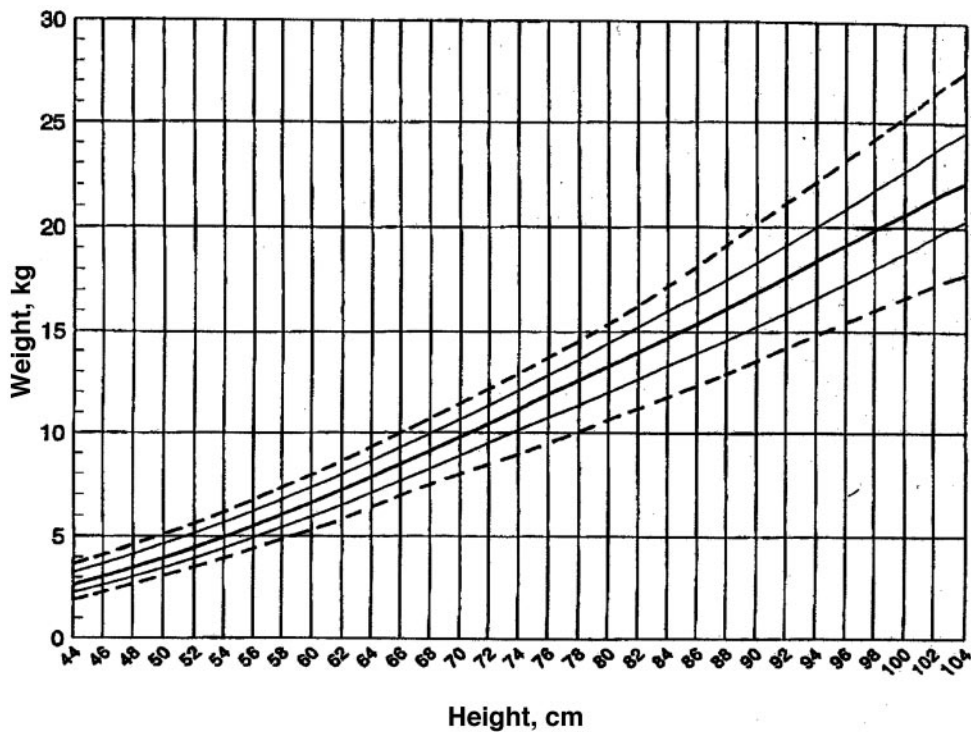


Fig 6. Height-by-weight standards in achondroplasia: males. (Reproduced with permission from *Am J Med Genet.* 1996;62:255-261.)

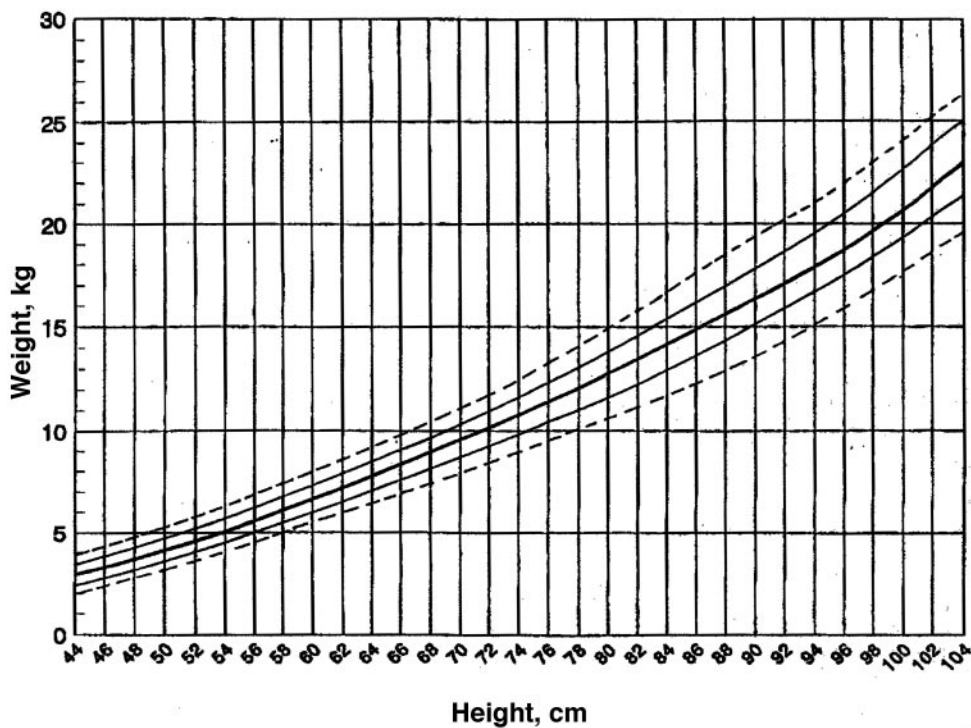


Fig 7. Height-by-weight standards in achondroplasia: females. (Reproduced with permission from *Am J Med Genet.* 1996;62:255-261.)

neural elements of interest; MRI provides such a direct assessment of the brainstem and upper cervical spinal cord, but no standards for estimation of foraminal size by MRI are available, and currently it cannot be performed routinely without sedation. Rapid development of imaging technology suggests that alternative methods may become appropriate in the future.

- If severe problems are found (eg, marked abnormality by neurologic examination, such as profound hypotonia or sustained ankle clonus; markedly diminished foramen magnum size compared with achondroplasia standard; substantial deformation of the upper cervical spinal cord; hypoxemic episodes with minimal oxygen saturations below 85%³¹), referral to a neurosur-

geon or other physician skilled and experienced in the care and treatment of neurologic problems in children with achondroplasia should be initiated.³⁴

- Hydrocephalus is a lifelong risk but is most likely to develop during the first 2 years.³⁵ OFC should be monitored carefully during this time. If the OFC is large or crosses percentiles on the achondroplasia-specific head-circumference chart, it is appropriate to refer the infant to a pediatric neurologist or pediatric neurosurgeon. Baseline CT or MRI (performed in conjunction with imaging of the craniocervical junction) is valuable if there is concern about possible hydrocephalus. Repeating neuroimaging to assess change in ventricular size should be considered if there is acceleration of head growth compared with achondroplasia standards or if other signs like bulging, hard fontanelle, or symptoms of unusual lethargy or intractable irritability develop. Both ventriculomegaly and excessive extra-axial fluid are common benign accompaniments of achondroplasia³⁵ and should not be misinterpreted as indicative of need for shunt placement. Neural ultrasonography may be used to follow these clinical findings.
- Restrictive pulmonary disease occurs in less than 5% of children with achondroplasia who are younger than 3 years.³⁶ Living at high elevation markedly increases the risk that restrictive problems will develop. If there are signs of respiratory distress or evidence of poor weight gain despite adequate caloric intake, pulse oximetry (during feeding, when crying, and at rest) should be considered to monitor oxygenation.
- Most infants with achondroplasia develop a thoracolumbar kyphosis. More severe kyphosis is associated with unsupported sitting before there is adequate trunk muscle strength.^{37,38} Parents should be counseled to avoid unsupported sitting and to avoid devices that cause curved sitting or "C sitting," such as "umbrella-style" strollers and soft canvas seats during the first year of life. Use of feeder seats for upright positioning should be recommended. If severe kyphosis appears to be developing, consider a pediatric orthopedic surgical assessment to determine if bracing is needed.³⁸
- The common complication of spinal stenosis rarely occurs in childhood but manifests in older individuals with numbness, weakness, and altered deep tendon reflexes.³⁰ Severe thoracolumbar kyphosis is one mechanism that can give rise to spinal stenosis. It is for this reason that unsupported sitting before there is adequate trunk muscle strength is discouraged.
- Anesthesia risk:³⁹ if an individual with achondroplasia needs to have anesthesia and surgery, the following should be considered:
 - (a) Care must be taken in manipulation of the neck, because uncontrolled neck movement (as may occur with intubation) could lead to unintentional spinal cord compression sec-

ondary to constriction of the foramen magnum.

- (b) Care should be taken to ensure that medication dosages are appropriate for size.
 - (c) Access to veins is sometimes difficult because of lack of full extension at the elbow.
 - (d) Generally, spinal anesthesia should be avoided, particularly when there is kyphosis or severe lumbar lordosis, because of limited space within the spinal canal.
 - (e) General anesthesia should be strongly considered for cesarean delivery for pregnant women with achondroplasia (they will all require caesarian delivery because of contracted pelvis), because use of epidural anesthesia in these women requires special skill and expertise.^{40/P}
3. Discuss the potential psychosocial implications for both parent and child related to disproportionate short stature. Refer the affected individual or the parent of an affected individual to a support group such as Little People of America (also see "Resources for New Parents"). If parents do not wish to join a group, they may want to meet with or talk to other affected individuals or parents. Remind parents that most individuals with achondroplasia lead productive, independent lives.
 4. Discuss with the parents how to tell their family and friends about their child's growth problem.
 5. Supply the parents with educational books and pamphlets (see "Resources for New Parents").
 6. Discuss the realistic functional problems for affected individuals.
 7. Discuss individual resources for support, such as family, clergy, social workers, psychologists, and friends.
 8. Review the prenatal diagnosis and recurrence risks for subsequent pregnancies.

HEALTH SUPERVISION FROM 1 MONTH TO 1 YEAR OF AGE: INFANCY

Examination

1. For infants not diagnosed in the newborn period, arrange for neuroimaging and polysomnography at the time of diagnosis.
2. Assess growth and development in comparison only with children with achondroplasia (Figs 1–7).
3. Perform physical examination.
4. Review head growth on achondroplasia-specific head-circumference charts.
5. Refer the infant to a pediatric neurologist or pediatric neurosurgeon if head size is disproportionately large or crosses percentiles, if there are signs or symptoms of hydrocephalus, or if there are indicators of possible craniocervical junction compression, including excessively brisk reflexes, asymmetric reflexes, ankle clonus, extreme hypotonia, or early hand preference.^{10,35}
6. Consider repeating neuroimaging studies if there is acceleration of head growth, severe persisting hypotonia, or any signs of craniocervical junction compression.^{31–34} Growth of the foramen magnum may be compared with achondroplasia-specific standards.⁴¹

7. Check motor development and discuss development; note on the milestone charts for achondroplasia. Expect motor delay but not social or cognitive delay.^{12,13} Check for serous otitis media. Review risk at 6 to 12 months of age. Formal behavioral audiometric assessment should be completed at 9 to 12 months of age. Language delay may be present secondary to conductive hearing loss.
8. Continue to monitor for progression of kyphosis at the thoracolumbar junction. It is recommended that parents avoid carrying a child with achondroplasia in curled-up (C-sitting) positions. Certain types of child carriers, mechanical swings, jumpers, and umbrella-style strollers tend to increase risk for gibbus. Unsupported sitting should be avoided.^{37,38} Parents and therapists should be instructed to provide back support during the first year of life. If severe kyphosis appears to be developing, consider pediatric orthopedic surgical assessment to determine if bracing is needed.³⁸
9. Be aware that external rotation of the hips is commonly present and usually disappears spontaneously when the child begins to bear weight. This finding does not require bracing for the infant.

Anticipatory Guidance

1. Review the personal support available to the family.
2. Review contact with support groups.
3. Observe the emotional status of parents and intrafamily relationships.
4. Discuss early-intervention services and the importance of normal socializing experiences with other children.
5. Ask the parents whether they have educated their family members about achondroplasia; discuss sibling adjustment.
6. Review the increased risk of serous otitis media because of short eustachian tubes. Indicate that an ear examination is appropriate with any persistent or severe upper respiratory tract infection or when parents suspect that ear pain may be present.
7. Advise parents to avoid infant carriers that curl up the infant. This does not apply to car safety seats, which should always be used during automobile travel. A rear-facing car safety seat should be used to the highest weight allowed by a convertible seat (25–30 lb).
8. Discuss filing for Supplemental Security Income benefits as appropriate.

HEALTH SUPERVISION FROM 1 TO 5 YEARS OF AGE: EARLY CHILDHOOD

Examination

1. Assess the child's growth and development as plotted on the achondroplasia growth charts.
2. Continue to follow rate of head growth on the achondroplasia-specific head-circumference charts.
3. Continue to watch for thoracolumbar gibbus (kyphosis). Discuss avoiding the use of walkers, jumpers, or backpack carriers. Any kyphosis present should disappear as the child begins to

bear weight. Lumbar lordosis usually develops but rarely requires specific intervention. Weight bearing and walking may occur late; however, they are expected by 2 to 2.5 years of age. When weight bearing begins, the external rotation of the hips should self-correct to a normal orientation within 6 months.

4. Anticipate some bowing of the legs. Many children will also have instability of the soft tissues surrounding the knee and internal tibial torsion. If positional deformity and instability leads to difficulty walking, a thrust at the knee (uncontrolled lateral or medial movement with weight bearing), or chronic pain, consult a pediatric orthopedist.¹⁴
5. Check the child's hips for hip-flexion contractures. Prescribe exercises that may decrease lumbar lordosis and hip-flexion contractures if indicated. Check the hips for external rotation. Refer the child to a pediatric orthopedist if necessary.
6. Screen hearing each year. If otologic history, hearing screening, or speech development raise concerns about hearing, formal audiologic assessment should be obtained.
7. Perform speech evaluation at no later than 2 years of age. If speech is delayed, conductive hearing loss attributable to chronic serous otitis media should be excluded.
8. Watch for obstructive sleep apnea secondary to smaller-than-average airway size plus physiologic adenoidal hypertrophy.^{42–44} Most children with achondroplasia snore. However, if obstructive apnea or disordered breathing in sleep is suspected (increased retraction, glottal stops, choking, intermittent breathing, apnea, deep compensatory sighs, secondary enuresis, recurrent night-time awakening or emesis), then additional pulmonary evaluation and polysomnography are indicated.
9. Be aware that gastroesophageal reflux may be more common in children with achondroplasia and may be more common in those with neurorespiratory complications.⁴⁵ If reflux is severe, in addition to usual treatments, consider referral to a pediatric specialist with experience in treating gastroesophageal reflux in infants and children.
10. Do not misinterpret greater-than-average sweating as indicative of serious medical problems; it is normal in children with achondroplasia.
11. In rare instances in which diagnosis of achondroplasia is delayed beyond 1 year of age, determine if neuroimaging is needed on the basis of clinical signs and symptoms.

Anticipatory Guidance

1. Consider adapting the home so that the child can become independent (eg, lower the light switches, use lever door handles and lever sink faucets, make the toilet accessible, and supply step stools) (see "Resources for Parents").
2. Determine if an occupational therapy consultation is needed.

3. Discuss adapting age-appropriate clothing with snapless, easy-opening fasteners and tuckable loops.
 4. Discuss adaptation of toys, especially tricycles, to accommodate short limbs.
 5. Discuss adaptation of toilets to allow comfortable, independent use, with an extended wand for wiping if needed.
 6. Discuss the use of a stool during sitting so that the child's feet are not hanging. Feet need support while the child is sitting at a desk, in a chair, or on the toilet. A cushion behind the child's back may be required for good posture and to prevent chronic back pain. Counsel parents for optimal protection to use a convertible rear-facing car safety seat to the highest weight and height allowed by the manufacturer of the seat.⁴⁶ A rear-facing seat provides the best support protection and positioning angle for a child with macrocephaly and skeletal dysplasia. Parents may benefit from suggestions for behavioral intervention to promote continuing the rear-facing position as long as possible.⁴⁷
 7. Review weight control and eating habits to avoid obesity, which often becomes a problem in mid-to late childhood.⁴⁸
 8. Discuss orthodontic bracing in the future and the possible need for early orthodontic assessment to consider palatal expansion.
 9. Encourage the family to develop activities in which the child can take part; avoid gymnastics, diving, trampolines, and collision sports.
 10. Discuss how to talk with the child and friends or family members about short stature.
 11. Encourage preschool attendance so that the child can learn to socialize in an age-appropriate way, and work with parents to prepare the teacher and the other children so that the child is not given unnecessary special privileges.
 12. Discuss toileting at school and special preparations needed by the school because of the child's short stature (see "Resources for Parents").
2. Discuss preparation of the school and teacher for a child with short stature (see "Resources for Parents").
 3. Prepare the child for others' questions and curiosity. Be sure the child can explain why he or she is short and can ask for help in an appropriate way. Children with achondroplasia usually are included in the regular education program.
 4. Suggest adaptive aids for the school to cope with heavy doors, high doorknobs, reaching for the blackboard, foot support, and a regular-sized desk. Also, be sure that the child can use the restroom independently (see "Resources for Parents").
 5. Counsel parents to use a child safety seat with a full harness to the highest weight allowed by the manufacturer of the seat and then to transition to the belt-positioning booster seat for optimal seat-belt positioning.⁴⁶
 6. Review socialization and foster independence.
 7. Discuss contact with support groups. They are especially valuable at this age.
 8. Consider obtaining an orthopedic evaluation when the child is approximately 5 years of age to make appropriate treatment plans if necessary.
 9. Emphasize correct posture and encourage the child to consciously decrease lumbar lordosis by "tucking the buttocks under." If lordosis is severe, consider physical therapy referral to teach lower abdominal muscle strengthening and pelvic rotation.
 10. Develop an activity program with acceptable activities such as swimming and biking. The child should avoid gymnastics and collision sports because of the potential for neurologic complications secondary to cervical spinal stenosis. If soccer is played, heading should be prohibited.
 11. Review orthodontic and speech status.

HEALTH SUPERVISION FROM 5 TO 13 YEARS OF AGE: LATE CHILDHOOD

Examination

1. Assess and review the child's growth, development, and social adaptation.
2. Plot measures on achondroplasia weight-by-height grids (Figs 6 and 7).
3. Review weight control.⁴⁸ The child may need to restrict food intake and eat less than an average-sized child eats.
4. Complete a general and neurologically oriented physical examination.
5. Check deep tendon reflexes yearly for asymmetry or increased reflexes that suggest spinal stenosis.
6. Continue to assess history for possible obstructive sleep apnea.
7. Test hearing each year.

Anticipatory Guidance

1. Determine school readiness.

HEALTH SUPERVISION FROM 13 TO 21 YEARS OR OLDER: ADOLESCENCE TO EARLY ADULTHOOD

Examination

1. Continue to record growth parameters.
2. Review weight control and diet.
3. Monitor for any signs or symptoms of nerve compression and check deep tendon reflexes, tone, and sensory findings, if indicated.
4. Continue to assess history for possible obstructive sleep apnea.

Anticipatory Guidance

1. Check on social adaptation.
2. Discuss the diagnosis with the adolescent to be sure that he or she has the vocabulary and the understanding of the genetic nature of achondroplasia.
3. Discuss contraception. The importance and use of contraception should be discussed with both males and females. Women with achondroplasia usually are fertile. Oral contraception should not be used long-term, because women with achondroplasia may have an increased risk of uterine fibroids, which may be aggravated by oral con-

traceptives. However, using a diaphragm is difficult because of the short arms in achondroplasia. Finding the appropriate long-term contraception may require consultation with a knowledgeable gynecologist. Women with achondroplasia often develop respiratory compromise late in pregnancy; thus, baseline respiratory function studies are recommended early in pregnancy. All pregnant women with achondroplasia will require caesarian delivery because of their small pelvic outlet. Because of spinal stenosis, spinal anesthesia is not recommended, and most women with achondroplasia should have general anesthesia for their cesarean delivery. If the partner of the woman with achondroplasia is of average stature, there will be a 50% risk of the infant having achondroplasia. If the partner also has short stature, the specific recurrence risk and the possibility of a severely affected infant must be determined so that the pregnancy and neonate can be managed properly. Prenatal diagnosis should be discussed to facilitate the management of the pregnancy for optimal outcome.¹⁸

4. Review orthodontic status.
5. Continue weight counseling.^{48,49}
6. Encourage the family and affected person to set career and life goals high and appropriate, as for other members of the family. Assist in adapting to an independent life and in obtaining a driver's license. Drivers usually require a vehicle that is adapted with pedal extenders; extenders that can be easily mounted and removed as needed are available. Families may wish to work with a driver-rehabilitation specialist who is qualified to assess the driver's transportation needs and who can provide them with a list of appropriate vehicle modifications. Names of qualified evaluators can be obtained by contacting a local rehabilitation center or the Association for Driver Rehabilitation Specialists (609-844-4433).⁵⁰ For most it will be necessary to provide a letter of justification for disabling of the air bag, because even with pedal extenders, marked arm foreshortening will preclude positioning at an appropriate distance from the air bag (10–12 in).⁴⁷ Individuals who want to have an air bag on-off switch must read an informational brochure and submit an official request to the National Highway Traffic Safety Administration (888-DASH-2-DOT [www.nhtsa.dot.gov]). Approval does not guarantee, however, that the request will be honored by a vehicle dealer.⁵¹
7. Discuss college, vocational planning and training, and other plans after high school.
8. Foster independence.
9. Continue to encourage participation in social activities and support groups. It is particularly useful during this age period.
10. Assist in transition to adult care.

COMMITTEE ON GENETICS, 2003–2004
G. Bradley Schaefer, MD, Chairperson
Marilyn J. Bull, MD
Joseph H. Hersh, MD

Celia I. Kaye, MD, PhD
Nancy J. Mendelsohn, MD
John B. Moeschler, MD
Howard M. Saal, MD
Chris Cunniff, MD
Past Committee Chairperson
Tracy L. Trotter, MD
Past Committee Member

CONTRIBUTORS

Judith G. Hall, OC, MD
Richard M. Pauli, MD, PhD

LIAISONS

James D. Goldberg, MD
American College of Obstetricians
and Gynecologists
James W. Hanson, MD
National Institute of Child Health
and Human Development
American College of Medical
Genetics
Michele Ann Lloyd-Puryear, MD,
PhD
Health Resources and Services
Administration
Sonja A. Rasmussen, MD
Centers for Disease Control and
Prevention

STAFF

Paul Spire

RESOURCES FOR PARENTS

Little People of America, Inc.
PO Box 65030
Lubbock, TX 79464-5030
www.lpaonline.org
Parent Coordinators: Grady and Mary Quick
4240 Oak Grove Dr
Carrollton, TX 75010

Books and Pamphlets

Kuklin S. *Thinking Big: The Story of a Young Dwarf*. New York, NY: Lothrop, Lee & Shepard Books; 1986

Campbell J, Dorren N. *It's a Whole New View: A Beginner's Guide for New Parents of a Child With Dwarfism*. Lubbock, TX: Little People America Inc; 1998 (available without charge from Little People of America at the address listed above)

To Celebrate: Understanding Developmental Differences in Young Children With Achondroplasia and Little People, Big Schools—Preparing the School for Your Young Child With Short Stature. Madison, WI: Midwest Regional Bone Dysplasia Clinic; 1997 (both available at cost from Midwest Regional Bone Dysplasia Clinic, University of Wisconsin, 1500 Highland Ave, Madison, WI 53705-2280)

Summary Chapters

Francomano CA. GeneReviews: achondroplasia. Available at: www.genetests.org. Accessed July 16, 2003

Pauli RM. Achondroplasia. In: Cassidy SB, Allanson JE, eds. *Management of Genetic Syndromes*. New York, NY: Wiley-Liss; 2001:9–32

REFERENCES

- American Academy of Pediatrics, Committee on Genetics. Health supervision for children with achondroplasia. *Pediatrics*. 1995;95:443–451
- Horton WA. Fibroblast growth factor receptor 3 and the human chondrodysplasias. *Curr Opin Pediatr*. 1997;9:437–442
- Pauli RM. Achondroplasia. In: Cassidy SB, Allanson JE, eds. *Management of Genetic Syndromes*. New York, NY: Wiley-Liss; 2001:9–32
- Orioli IM, Castilla EE, Scarano G, Mastroiacovo P. Effect of paternal age in achondroplasia, thanatophoric dysplasia, and osteogenesis imperfecta. *Am J Med Genet*. 1995;59:209–217
- Shiang R, Thompson LM, Zhu YZ, et al. Mutations in the transmembrane domain of FGFR3 cause the most common genetic form of dwarfism, achondroplasia. *Cell*. 1994;78:335–342
- Rousseau F, Bonaventure J, Legeai-Mallet L, et al. Mutations in the gene encoding fibroblast growth factor receptor-3 in achondroplasia. *Nature*. 1994;371:252–254
- Bellus GA, Hefferon TW, Ortiz de Luna RI, et al. Achondroplasia is defined by recurrent G380R mutations in FGFR3. *Am J Hum Genet*. 1995;56:368–373
- Francomano CA. GeneReviews: achondroplasia. Available at: www.genetests.org. Accessed July 16, 2003
- Horton WA, Rotter JI, Rimoin DL, Scott CI, Hall JG. Standard growth curves for achondroplasia. *J Pediatr*. 1978;93:435–438
- Hecht JT, Butler IJ. Neurologic morbidity associated with achondroplasia. *J Child Neurol*. 1990;5:84–97
- Pyeritz RE, Sack GH Jr, Udvarhelyi GB. Thoracolumbosacral laminectomy in achondroplasia: long-term results in 22 patients. *Am J Med Genet*. 1987;28:433–444
- Todorov AB, Scott CI Jr, Warren AE, Leeper JD. Developmental screening tests in achondroplastic children. *Am J Med Genet*. 1981;9:19–23
- Fowler ES, Glinski LP, Reiser CA, Horton VK, Pauli RM. Biophysical basis for delayed and aberrant motor development in young children with achondroplasia. *J Dev Behav Pediatr*. 1997;18:143–150
- Kopits SE. Surgical intervention in achondroplasia. Correction of bow-leg deformity in achondroplasia. *Johns Hopkins Med J*. 1980;146:206–209
- Thompson NM, Hecht JT, Bohan TP, et al. Neuroanatomic and neuropsychological outcome in school-age children with achondroplasia. *Am J Med Genet*. 1999;88:145–153
- Modaff P, Horton VK, Pauli RM. Errors in the prenatal diagnosis of children with achondroplasia. *Prenat Diagn*. 1996;16:525–530
- Little People of America. Adopting a dwarf child. Available at: http://lpaonline.org/lpa_adoptions.html. Accessed July 16, 2003
- Allanson JE, Hall JG. Obstetrics and gynecologic problems in women with chondrodysplasias. *Obstet Gynecol*. 1986;67:74–78
- Flynn M, Pauli RM. Double heterozygosity and compound heterozygosity for disorders affecting bone growth. *Am J Med Genet*. 2003;121A:193–208
- Pauli RM, Conroy MM, Langer LO Jr, et al. Homozygous achondroplasia with survival beyond infancy. *Am J Med Genet*. 1983;16:459–473
- Mettler G, Fraser FC. Recurrence risk for sibs of children with “sporadic” achondroplasia. *Am J Med Genet*. 2000;90:250–251
- Henderson S, Sillence D, Loughlin J, Bennetts B, Sykes B. Germline and somatic mosaicism in achondroplasia. *J Med Genet*. 2000;37:956–958
- Sobetzko D, Braga S, Rudeberg A, Superti-Furga A. Achondroplasia with the FGFR3 1138g→a (G380R) mutation in two sibs sharing a 4p haplotype derived from their unaffected father. *J Med Genet*. 2000;37:958–959
- Kanaka-Gantenbein C. Present status of the use of growth hormone in short children with bone diseases (diseases of the skeleton). *J Pediatr Endocrinol Metab*. 2001;14:17–26
- Seino Y, Yamanaka Y, Shinohara M, et al. Growth hormone therapy in achondroplasia. *Horm Res*. 2000;53(suppl 3):53–56
- Villarubias JM, Ginebreda I, Jimeno E. Lengthening of the lower limbs and correction of lumbar hyperlordosis in achondroplasia. *Clin Orthop*. 1990;250:143–149
- Aldegheri R, Dall’Oca C. Limb lengthening in short stature patients. *J Pediatr Orthop B*. 2001;10:238–247
- Pauli RM, Scott CI, Wassman ER Jr, et al. Apnea and sudden unexpected death in infants with achondroplasia. *J Pediatr*. 1984;104:342–348
- Reid CS, Peyeritz RE, Kopits SE, et al. Cervicomedullary compression in young patients with achondroplasia: value of comprehensive neurologic and respiratory evaluation. *J Pediatr*. 1987;110:522–530
- Hecht JT, Butler IJ, Scott CI Jr. Long-term neurological sequelae in achondroplasia. *Eur J Pediatr*. 1984;143:58–60
- Pauli RM, Horton VK, Glinski LP, Reiser CA. Prospective assessment of risks for cervicomedullary-junction compression in infants with achondroplasia. *Am J Hum Genet*. 1995;56:732–744
- Bruhl K, Stoeter P, Wietek B, et al. Cerebral spinal fluid flow, venous drainage and spinal cord compression in achondroplastic children: impact of magnetic resonance findings for decompressive surgery at the cranio-cervical junction. *Eur J Pediatr*. 2001;160:10–20
- Rollins N, Booth T, Shapiro K. The use of gated cine phase contrast and MR venography in achondroplasia. *Childs Nerv Syst*. 2000;16:569–575; discussion 575–577
- Keiper GL Jr, Koch B, Crone KR. Achondroplasia and cervicomedullary compression: prospective evaluation and surgical treatment. *Pediatr Neurosurg*. 1999;31:78–83
- Steinbok P, Hall J, Flodmark O. Hydrocephalus in achondroplasia: the possible role of intracranial venous hypertension. *J Neurosurg*. 1989;71:42–48
- Stokes DC, Phillips JA, Leonard CO, et al. Respiratory complications of achondroplasia. *J Pediatr*. 1983;102:534–541
- Hall JG. Kyphosis in achondroplasia: probably preventable. *J Pediatr*. 1988;112:166–167
- Pauli RM, Breed A, Horton VK, Glinski LP, Reiser CA. Prevention of fixed, angular kyphosis in achondroplasia. *J Pediatr Orthop*. 1997;17:726–733
- Berkowitz ID, Raja SN, Bender KS, Kopits SE. Dwarfs: pathophysiology and anesthetic implications. *Anesthesiology*. 1990;73:739–759
- Morrow MJ, Black IH. Epidural anaesthesia for caesarean section in an achondroplastic dwarf. *Br J Anaesth*. 1998;81:619–621
- Hecht JT, Horton WA, Reid CS, Peyeritz RE, Chakraborty R. Growth of the foramen magnum in achondroplasia. *Am J Med Genet*. 1989;32:528–535
- Waters KA, Everett F, Sillence DO, Fagan ER, Sullivan CE. Treatment of obstructive sleep apnea in achondroplasia: Evaluation of sleep, breathing, and somatosensory-evoked potentials. *Am J Med Genet*. 1995;59:460–466
- Zucconi M, Weber G, Castronovo V, et al. Sleep and upper airway obstruction in children with achondroplasia. *J Pediatr*. 1996;129:743–749
- Mogayzel PJ Jr, Carroll JL, Loughlin GM, Hurko O, Francomano CA, Marcus CL. Sleep-disordered breathing in children with achondroplasia. *J Pediatr*. 1998;132:667–671
- Tasker RC, Dundas I, Laverty A, Fletcher M, Lane R, Stocks J. Distinct patterns of respiratory difficulty in young children with achondroplasia: a clinical, sleep, and lung function study. *Arch Dis Child*. 1998;79:99–108
- American Academy of Pediatrics, Committee on Injury, Violence, and Poison Prevention. Selecting and using the most appropriate car safety seats for growing children: guidelines for counseling parents. *Pediatrics*. 2002;109:550–553
- Automotive Safety Program. *Transporting Children With Special Health Care Needs: Training and Resource Manual*. Indianapolis, IN: Automotive Safety Program; 2000:13–14
- Hecht JT, Hood OJ, Schwartz RJ, Hennessey JC, Bernhardt BA, Horton WA. Obesity in achondroplasia. *Am J Med Genet*. 1988;31:597–602
- Hunter AG, Hecht JT, Scott CI Jr. Standard weight for height curves in achondroplasia. *Am J Med Genet*. 1996;62:255–261
- National Highway Traffic Safety Administration. *Adapting Motor Vehicles for People With Disabilities*. Washington, DC: National Highway Traffic Safety Administration; 2000. Available at: www.nhtsa.dot.gov/cars/rules/adaptive/brochure/brochure.html. Accessed February 3, 2004
- National Highway Traffic Safety Administration. *Standardized Child Passenger Safety Training Program: Instructor Guide*. Washington, DC: National Highway Traffic Safety Administration; 2000:F-22–F-23

All clinical reports from the American Academy of Pediatrics automatically expire 5 years after publication unless reaffirmed, revised, or retired at or before that time.

Health Supervision for Children With Achondroplasia

Tracy L. Trotter and Judith G. Hall

Pediatrics 2005;116;771

DOI: 10.1542/peds.2005-1440

Updated Information & Services

including high resolution figures, can be found at:
<http://pediatrics.aappublications.org/content/116/3/771>

References

This article cites 44 articles, 5 of which you can access for free at:
<http://pediatrics.aappublications.org/content/116/3/771.full#ref-list-1>

Subspecialty Collections

This article, along with others on similar topics, appears in the following collection(s):

Current Policy

http://classic.pediatrics.aappublications.org/cgi/collection/current_policy

Council on Genetics

http://classic.pediatrics.aappublications.org/cgi/collection/committee_on_genetics

Genetics

http://classic.pediatrics.aappublications.org/cgi/collection/genetics_sub

Permissions & Licensing

Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
<https://shop.aap.org/licensing-permissions/>

Reprints

Information about ordering reprints can be found online:
<http://classic.pediatrics.aappublications.org/content/reprints>

Pediatrics is the official journal of the American Academy of Pediatrics. A monthly publication, it has been published continuously since . Pediatrics is owned, published, and trademarked by the American Academy of Pediatrics, 141 Northwest Point Boulevard, Elk Grove Village, Illinois, 60007. Copyright © 2005 by the American Academy of Pediatrics. All rights reserved. Print ISSN: .

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™



ERRATA

An error appeared in the Experience and Reason—Briefly Recorded by Tan et al, titled “Isolated Sulfite Oxidase Deficiency: A Case Report With a Novel Mutation and Review of the Literature” that was published in the September 2005 issue of *Pediatrics* (2005;116:757–766). On page 758, the last line in column 2, the authors wrote: “The patient was homozygous for a novel 4-base pair deletion (c.2037-2040delTAGA) in exon 3 that led to a frameshift and resulted in a prematurely truncated protein.” This sentence should have read as follows: “The patient was homozygous for a novel 4-base pair deletion (c.1142-1145delTAGA) in exon 3 that led to a frameshift and resulted in a prematurely truncated protein.” The same error appears on page 763, Table 1, column 22 in the Mutations in SUOX row. The table currently reads: “c2037-2040delTAGA.” It should read: “c.1142-1145delTAGA.” The authors state that to the best of their knowledge, this remains a new, previously unreported mutation, and this error in nomenclature does not alter the substance nor the importance of the publication in any way.

doi:10.1542/peds.2005-2563

An error occurred in the AAP clinical report by Trotter et al, titled “Health Supervision for Children With Achondroplasia” that was published in the September 2005 issue of *Pediatrics* (2005;116:771–783). Figures 1, 2, 4, and 5 were credited incorrectly. The credit for figures 1, 2, 4, and 5 should read: Saul RA, Seaver LH, Sweet KM, Geer JS, Phelan MC, Mills CM. *Growth References: Third Trimester to Adulthood*. 2nd ed. Greenwood, SC: Greenwood Genetic Center; 1998: 107–110.

doi:10.1542/peds.2005-2461

An error appeared in the commentary by Cherry titled “Pertussis Vaccines for Adolescents and Adults” that was published in the September 2005 issue of *Pediatrics* (2005;116:755–756). On page 756, in Table 1 in the column under Boostrix for the entry 2-Phenoxyethanol, the 2.5 mg should be removed and replaced by a dash (—).

TABLE 1. Composition per 0.5-mL Dose of Adacel and Boostrix

Antigen	Adacel	Boostrix
Diphtheria toxoid	2 Lf	2.5 Lf
Tetanus toxoid	5 Lf	5 Lf
Pertussis toxin toxoid	2.5 µg	8 µg
Filamentous hemagglutinin	5 µg	8 µg
Pertactin	3 µg	2.5 µg
Fimbriae 2/3	5 µg	—
Aluminum	0.3 mg	0.3 mg
2-Phenoxyethanol	0.6%	—

Lf indicates flocculation units.

doi:10.1542/peds.2005-2447

PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Health Supervision for Children With Achondroplasia

Tracy L. Trotter and Judith G. Hall

Pediatrics 2005;116:771

DOI: 10.1542/peds.2005-1440

The online version of this article, along with updated information and services, is located on the World Wide Web at:

<http://pediatrics.aappublications.org/content/116/3/771>

An erratum has been published regarding this article. Please see the attached page for:

<http://pediatrics.aappublications.org/content/116/6/1615.2.full.pdf>

Pediatrics is the official journal of the American Academy of Pediatrics. A monthly publication, it has been published continuously since . Pediatrics is owned, published, and trademarked by the American Academy of Pediatrics, 141 Northwest Point Boulevard, Elk Grove Village, Illinois, 60007. Copyright © 2005 by the American Academy of Pediatrics. All rights reserved. Print ISSN:

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

