Health Supervision for Children With Achondroplasia
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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, craniofacial 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.

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 sacrosciatic 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, craniofacial 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-
ductive lives. 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
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. 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.
4. Review the natural history and manifestations of achondroplasia, including variability.\(^3\)

5. 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.

6. 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.

7. 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.\(^17\)

8. 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.

9. When both parents are of disproportionate short stature, assess the possibility of double heterozygosity\(^19\) or homozygosity for achondroplasia. Some forms of double heterozygosity lead to life-threatening problems\(^19\); infants with homozygous achondroplasia usually are stillborn or die shortly after birth.\(^20\)

**HEALTH SUPERVISION FROM BIRTH TO 1 MONTH OF AGE: NEWBORNS**

**Examination**

1. 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).

2. 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.

3. The OFC should be measured at every pediatric contact during the first year (Figs 4 and 5).
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<th>TABLE 1. Achondroplasia Guidelines for Health Supervision</th>
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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.
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
persons with achondroplasia is approximately 4 ft.\textsuperscript{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.\textsuperscript{28,29} This seems to result from central apnea arising secondary to compression of arteries at the level of the foramen magnum.\textsuperscript{28} In addition, the universally small foramen magnum may result in a high cervical myelopathy.\textsuperscript{30,31} However, with appropriate assessment and intervention, both risks can be minimized.\textsuperscript{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.\textsuperscript{31} Neuroimaging can be by computed tomography (CT) with thin cuts and bone windows\textsuperscript{31} or magnetic resonance imaging (MRI),\textsuperscript{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 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.)]
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%\(^3\)), referral to a neurosur-
Anesthesia risk:39 if an individual 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 secondary 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 cesarean 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.\textsuperscript{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.\textsuperscript{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.\textsuperscript{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.\textsuperscript{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.\textsuperscript{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.\textsuperscript{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, 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. 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.
7. Review weight control and eating habits to avoid obesity, which often becomes a problem in mid- to late childhood.
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”).

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. 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.
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 seatbelt positioning.
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 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 cesarean 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.

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Books and Pamphlets
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
REFERENCES


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.
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

<table>
<thead>
<tr>
<th>Antigen</th>
<th>Adacel</th>
<th>Boostrix</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diphtheria toxoid</td>
<td>2 Lf</td>
<td>2.5 Lf</td>
</tr>
<tr>
<td>Tetanus toxoid</td>
<td>5 Lf</td>
<td>5 Lf</td>
</tr>
<tr>
<td>Pertussis toxin toxoid</td>
<td>2.5 μg</td>
<td>8 μg</td>
</tr>
<tr>
<td>Filamentous hemagglutinin</td>
<td>5 μg</td>
<td>8 μg</td>
</tr>
<tr>
<td>Pertactin</td>
<td>3 μg</td>
<td>2.5 μg</td>
</tr>
<tr>
<td>Fimbriae 2/3</td>
<td>5 μg</td>
<td>—</td>
</tr>
<tr>
<td>Aluminum</td>
<td>0.3 mg</td>
<td>0.3 mg</td>
</tr>
<tr>
<td>2-Phenoxyethanol</td>
<td>0.6%</td>
<td>—</td>
</tr>
</tbody>
</table>

Lf indicates flocculation units.

doi:10.1542/peds.2005-2447