HUMAN GROWTH FOUNDATION

Achondroplasia

An introduction to achondroplasia for patients, their families and friends, and healthcare providers

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Skeletal Dysplasia: Definition and Background

- ~250 Skeletal Dysplasia Diagnoses are recognized today
- Achondroplasia is the most common short stature dysplasia
- ~ 350,000 people have achondroplasia in the world

What is a skeletal dysplasia?

The term 'skeletal dysplasia' means a condition of abnormal bone formation. There are over 250 different skeletal dysplasia diagnoses recognized today. Each dysplasia diagnosis affects the spine, extremities and/or the skull in different ways, but a common feature of nearly all the diagnoses is short stature. Achondroplasia is the most common short stature skeletal dysplasia.

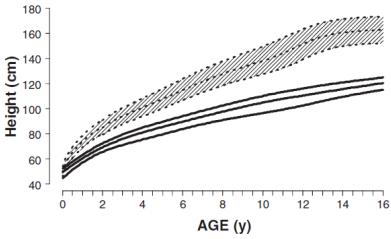
What is achondroplasia?

Achondroplasia is the most common short stature skeletal dysplasia. The birth prevalence is approximately 1 in 20,000 to 1 in 30,000 live births. Therefore, there are 15,000+ individuals with achondroplasia in the US and 350,000+ globally. Males and females are equally affected, and it occurs uniformly around the world. Approximately 80% of all people with achondroplasia are the first person in their family to have the diagnosis. Achondroplasia is a dominant condition, so each affected individual has a 50% chance to pass this condition to each child.

Common Physical Features in Everyone with Achondroplasia

Long Trunk, Short Limbs
The source of short stature in achondroplasia is primarily in the limbs. The trunk is relatively similar to average stature.

Short Stature
Achondroplasia length/height curves deviate from average stature shortly after birth. Final adult male height is ~ 4'2" and female is ~4'.

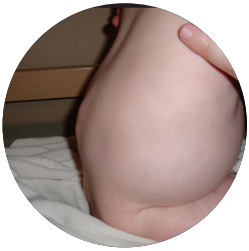


Macrocephaly Macrocephaly means 'large head'. In most people with achondroplasia, macrocephaly is normal because the brain is larger and there is increased fluid in the ventricles and around the brain. Macrocephaly is abnormal when it causes increased intracranial pressure which is treated with surgery.

Head circumference should be measured at all healthcare encounters from birth until school age and plotted on achondroplasia-specific curves.

**See curves page

Mid-Face Hypoplasia
The middle section of the face (i.e. cheeks and nose) is recessed (set back further) due to abnormal bone growth in the face and skull. This may contribute to obstructive sleep apnea.



Rhizomelia (short proximal limbs), decreased elbow extension, trident hands (splayed fingers) These are normal limb features in patients with achondroplasia, caused by differences in skeletal growth.
**Collecting blood from a vein in the arm may be challenging due to decreased elbow extension. Try rotating the arm laterally or use other sites such as feet, hands or scalp in infants.

Thoracolumbar kyphosis
This is posterior curvature of the mid to lower spine. It is noticeable while young children sit and usually resolves when they can stand. Kyphosis should be monitored closely in childhood. The goal is to prevent kyphosis from becoming permanent. **See discussion in Infancy and Early Childhood pages

Genu Varus (bowed legs) **and tibial bowing** (outward bowing of legs) These result from the misshapen metaphyses (end of the long bones) in achondroplasia. Surgery may be performed to realign the legs if pain and decreased function is severe.

Achondroplasia Over the Lifespan: PRENATAL

For **Average Stature** Mothers Having a Child with Achondroplasia

Ultrasound:
The prenatal diagnosis of achondroplasia by ultrasound is not usually suspected until the third trimester when limb shortening becomes more noticeable. Limb shortening, macrocephaly and mid-face hypoplasia—all features of achondroplasia—may be subtle. In an average stature mother, the diagnosis of achondroplasia in her infant is often discussed for the first time after birth.

Non-invasive prenatal testing (NIPT): There are a few labs in the US performing testing for dominant conditions from fetal DNA collected from a maternal blood sample. The diagnosis of achondroplasia may be detected with this version of NIPT. Prenatal carrier screening is a different NIPT option, focused on recessive conditions. Since achondroplasia is a dominant condition, it is not detected by prenatal carrier screening.

Invasive prenatal testing options:
An average stature mother may pursue testing for genetic conditions from a sample of fetal DNA collected by chorionic villus sampling (around 9-11 weeks gestation) or amniocentesis (around 16-20 weeks gestation). Genetic testing of the sample may include chromosome differences, a single gene or a group of genes. Remember, the skeletal features of achondroplasia may not be apparent until the third trimester, so invasive prenatal testing may not have included FGFR3 (the gene that causes achondroplasia).

A C-section may be necessary for an average stature mother of a child with achondroplasia because of the large head size.

For **Mothers** or **Partner with Achondroplasia** Having a Child with Achondroplasia

Achondroplasia in the prenatal period by ultrasound: A person with achondroplasia has a 50% chance to pass the condition on to each child. A Maternal-Fetal Medicine expert or ultrasonographer aware of parental short stature may detect subtle skeletal differences earlier in gestation. Otherwise the features may be noticed for the first time after birth.

Both parents have a skeletal dysplasia: If the mother with achondroplasia has a partner with a short stature dysplasia, the risk and health expectations of the child are different. Genetic counseling and a medical evaluation are recommended to confirm the diagnoses of both parents and discuss the risk of lethality in infants inheriting both parental skeletal dysplasias.

Invasive prenatal testing options: This may be pursued if the mother/parents are concerned the fetus inherited one or both of their dysplasias. Inheriting both dysplasias may be lethal. A Maternal-Fetal Medicine expert or genetic counselor will assist with testing.

Preimplantation genetic diagnosis for a specific dysplasia: If both parents have a short stature dysplasia confirmed by genetic testing, egg and sperm may be collected from each, fertilized in the laboratory by in vitro fertilization, and a single cell from the fertilized zygote tested for the presence of 1 or 2 known dysplasia mutations. Typically the zygotes without any known mutation are implanted.

Pregnant women with a dysplasia: These women may deliver a couple weeks early due to limited trunk size to support maternal respiration. She should expect to deliver by a C-section. Pre-pregnancy assessment of the spine for suitability of epidural anesthesia for delivery is recommended.

Achondroplasia Over the Lifespan: INFANCY & EARLY CHILDHOOD

HIGHLIGHTS:

1. Risk of compression of upper spinal cord and brain stem: To assess for compression, all infants and young children with achondroplasia should have regular physical examinations with a neurologic exam, assessment of development and growth, and ideally an MRI of the neck and brain and a sleep study.

2. Developmental Milestones: Developmental motor milestones are different for infants and young children with achondroplasia than their age-matched peers due to the large head and decreased tone. Sitting up, for example, is usually always later than average stature. **See charts on PAGE Overall, cognitive and speech development should be the same as peers.

3. Noisy Breathing: It is normal for infants and young children with achondroplasia to have noisy breathing with mild congestion that improves with saline wash and bulb syringe. Long pauses, struggling to breathe, blue color changes around the mouth and lips, or difficulty eating and breathing are NOT normal—seek medical attention immediately if present.

4. Avoid prolonged unsupported sitting or holding: The goal is to avoid kyphosis in infants and young children with achondroplasia. Until he/she can sit up straight (i.e. without kyphosis) with good head control, stand and walk, encouraging 'belly and back' time instead is recommended.

5. Use firm, high-back chair with incline: Limited use of a firm reclining high-chair may be used as long as the baby is supervised and not left to sit for a long period of time. **See page X for suggestions on a high chair.

6. No child should sleep in a car seat unattended: This is especially important in infants and young children with achondroplasia in whom the head is large and heavy enough that the neck could bend and obstruct (close) the airway.

CAR SEAT INFORMATION

WARNING: This car seat information is supplied by a Certified Child Passenger Safety Technician (CCPST). The National Highway Traffic Safety Administration certifies CCPSTs. Refer to the car seat manufacturer's instructions and your state guidelines to install your car seat properly. Safe Kids Worldwide has a website (safekids.com) with the latest car seat safety information. *The authors of this document are not responsible for the safe installation of your car seat.*

Car Seats for a Child with Achondroplasia:

Rear-facing car seats are the safest for all children. Children should be rear-facing as long as they fit in their car seat and are under the maximum weight standard for that seat. A child with achondroplasia may fit well in a rear-facing car seat for 4 years or longer because of his/her shorter legs.

Car Bed Option for A Child with Achondroplasia:

In some cases, a car bed may be a good option when taking a child with achondroplasia home from the hospital. The Cosco Dream Ride car bed can be used for babies who are 5 to 20 pounds, with a maximum height of 26 inches.

Reclining car seat avoids back curvature

A child with achondroplasia will sit up later than average height peers. A reclining car seat is recommended to avoid 'slouching' while sitting in the car seat. A car seat with firm back support, positioned at the maximum reclining angle is ideal.

Transition from rear-facing car seat to booster seat:

Ideally a child will remain rear-facing for as long as possible. There are state-specific laws for height and weight at which a child can transition to forward-facing car seats (e.g. In Maryland, 57 inches and 80 pounds). Please check your state-specific guidelines.

Psychosocial Pearl

Sometimes a child with achondroplasia may never attain the recommended height and weight to stop using a car seat or booster seat. Each parent and child need to decide together what is the best age for these transitions. As average height peers grow physically out of their car seats, the person with achondroplasia may mentally grow out of theirs. It is important to keep kids safe, but also maintain age-appropriate behavior and self-esteem. Cultivating open communication is key to managing this situation.

Booster car seat:

Since people with achondroplasia have torsos around the same height as their average stature peers, the seat belt should cross at the same place on their bodies.

Achondroplasia Over the Lifespan: SCHOOL AGE

HIGHLIGHTS:

1. 504 plan: Seek out a 504 plan at school for adaptations. The public school setting, by law, must be accessible to every student regardless of physical disabilities. **See PAGES for more information.

2. Developmental milestones: By school age, developmental milestones should be on par with age-matched peers. If there is speech delay, check hearing for deficits related to chronic middle ear fluid/infection. If there are difficulties in academic achievement, evaluate the child with achondroplasia *as you would any child in school.*

3. Sleep disordered breathing: If there is excessive daytime sleepiness, inability to concentrate in school, heavy snoring or observed apnea, consider a sleep study to assess for sleep disordered breathing.

4. Unsupported sitting: Avoid prolonged unsupported sitting. Encourage step stool for feet or bolster behind back while sitting in a desk.

5. Discussion with teacher(s) about child with short stature: Prepare the school and teacher for a child with short stature. The child needs to be involved in the discussion about communication with and awareness of classmates and school faculty.

6. Elevator speech Prepare the child to answerquestions about his/her height from curious peers. **See “the elevator speech” on page X.

7. Sports and physical activity: Participatioin sports and physical activity is encouraged for everyone with short stature for socialization and weight management.

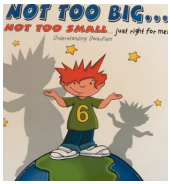
8. Signs of neurologic problems: Assess for pain, numbness or tingling int he extremities and seek evaluation if it is hindering daily activities.

Dwarfism Awareness in School

In elementary school, it is helpful to educate the other students about dwarfism in the first couple of weeks of the new school year. This is especially important in kindergarten where most have never encountered a person with achondroplasia. The parent can request to speak to the kindergarten classes about achondroplasia and ways their child may modify activities to participate in classroom events. The student should always be included in this decision and know that this is happening in his school (but not be forced to participate if he does not want to).

Psychosocial Pearl

Listen and include. Every child will have their own opinion and comfort level regarding adaptations for school. He or she should be included in decisions regarding seating, dwarfism awareness materials, and physical education adaptations. Some children are more sensitive about appearing "different", so a modified desk that looks exactly the same as everyone else's may be more important than the teacher or parent would imagine.



Reading a book to the class to open up the conversation is a great way to explain dwarfism, acceptance, and similarities/differences. After the book, the students can have a chance to ask the questions they've been trying to ask in the hallway, when they saw the child with achondroplasia for the first time. Opening up this communication helps alleviate staring and pointing throughout the year.

SCHOOL RESOURCES

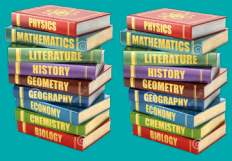
504 and Individual Education Plans (IEP)

A child with achondroplasia qualifies for a Section 504 Plan at a public school. A Section 504 Plan is the result of a federal civil rights law, which falls under the Section 504 Rehabilitation Act of 1973. A 504 Plan is a blueprint for all public schools to provide support and remove barriers for a student with a disability. Since achondroplasia is a disability recognized by the Americans with Disabilities Act (ADA), there is no other diagnosis necessary to obtain a 504 plan for a child in a public school.

In the bathroom area, stools should be in place so that bathroom sinks/soap, toilets, and paper towels can be reached. Stools should also be used in the classrooms and anywhere necessary to encourage the child to be as independent as his/her peers. At times, doors may be too heavy or doorknobs too high, and may need to be modified so that the child can access all school rooms needed.

Seating can be modified with stools and cushions for back support. As children get older, they may not want to have obvious modifications because they do not want to look different than their peers. The student should be involved in their 504 process and agree to the modifications.

Heavy backpacks are discouraged. There are several ways to alleviate this issue; two sets of books—one at home and one at school, books online, and rolling backpacks. In middle and high school, scheduling classes together (i.e. on one level of school) and decreasing the distance between classes are options.



Physical education activities and fitness requirements can also be adapted to include the child with achondroplasia with their classmates. The main reason to ensure a child with achondroplasia has a 504 plan is to make their learning environment as assessable as possible.

An Individual Education Plan (IEP) is different than a 504 plan. An IEP is a federal special education law under the Individuals with Disabilities Education Act (IDEA). It assures individualized special education and related services are provided to meet a child's unique needs. A child with achondroplasia would need an additional reason (i.e. speech delay, learning disability) to qualify for an IEP. These issues may occur in any child-including those with achondroplasia—but are not expected just because of that diagnosis.

Stairs at School

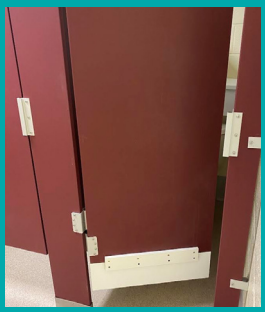
Preschool through Elementary In early school years, children are usually very excited to go to a new location, and sometimes in the excitement, there can be friendly shoving in the halls and classrooms. The student with achondroplasia should be at the back of the line when walking downstairs with his peers. Walking at the front of the line could be unsafe if he is walking downstairs slower than this friends.

Middle and High School Years The staircases tend to be crowded, and the student with achondroplasia should have the option to take the elevator if she/he chooses. In some 504 plans, the student is allowed to leave 5 minutes early from class to have more time in a less crowded hallway to reach her next class.

Use of Scooters

If a child with achondroplasia is having a tough time keeping up with her friends while walking, a small, manual scooter (i.e. Razor) can be approved in the 504 or IEP plan for use in the school. With a manual scooter, the student is getting exercise and can navigate the hall and school more quickly.

Student with achondroplasia accessing i his schoolbus independently



Bathroom stall modified to ensure more privacy for student with dwarfism

Achondroplasia Over the Lifespan: TEENS

HIGHLIGHTS:

1. 504 plan: Seek out 504 plan at school for adaptations. public school setting, by law, must be accessible to every student regardless of physical disabilities. **See pages X for more information.

2. School Performance: If there are difficulties in academic achievement, evaluate the child with achondroplasia as any child in school. This may include academic testing as well as audiology to ensure hearing is normal.

3. Sleep disordered breathing: If there is excessive daytime sleepiness, inability to concentrate in school, heavy snoring or observed apnea, consider a sleep study to assess for sleep disordered breathing.

4. Unsupported sitting posture: Avoid prolonged unsupported sitting. Encourage step stool for feet or bolster behind back while sitting in a desk.

5. Bullying: Inquire about bullying in the school setting. **See pages XX.

6. Signs of neurologic problems: Assess for pain, numbness or tingling in the extremities and seek evaluation if it is hindering daily activities. Evaluate whether an activity that a person used to be able to do remains easy. For example, if the teen could walk the mall without issue months ago, but now has to squat or complains of pain, it might be time for a check up with the skeletal dysplasia team.

7. Weight management and Exercise in the context of sport selection: **See pages

8. Post high school plans: Discuss post-high school occupational, academic and life plans. People with achondroplasia historically have a variety of careers, ranging from doctors to writers to firefighters to plumbers. Consider which careers require heavy lifting or long periods of standing. Most of the time there are ways to adapt job settings for people with achondroplasia to have a wide array of choices in employment.

9. Driving: Discuss driving adaptation options (e.g. pedal extenders). Driving should not be an issue for people with achondroplasia. There are many types of pedal extensions that are available and are relatively easy to install, even in rental cars while traveling. Some people with achondroplasia choose to drive with hand controls, but most purchase pedal extensions.

10. Transitioning medical responsibility from parent to child: At Encourage teen to begin taking responsibility for overall medical care including non-achondroplasia care. Teens and young adults may think all of their medical care has been covered if they check in with their dwarfism specialist (and vice versa). Routine medical care, such as physicals, labs, dermatology visits, etc. should be as consistent as average height peers. Having achondroplasia does not give a person a 'pass' on other medical issues, unfortunately, so it's important to stay on top of routine care.

9. Chance to pass achodroplasia to future childre: Discuss 50% chance to pass on achondroplasia to each future child and tha recurrence risks change if their future partner also has a short stature. ****Please see pages

10. Puberty and fertility: Please Normal pubertal development is expecte with fertility intact. ****Please see pages

PUBERTY, DATING & SOCIAL LIFE

Pubertal Development

Teens with achondroplasia have normal pubertal development, and experience puberty around the same age range as average stature children. When purchasing tampons, it is easier to use a slightly shorter tampon with an applicator, such as the Playtex brand. Fertility is the same as average stature peers, so talking about intimacy, birth control and sex education is advisable to do at the same age as average stature siblings.

Social Life and Dating

It may be difficult at times for a teen with achondroplasia in a typical high school setting to date his or her peers. It's important to acknowledge their feelings and encourage them to talk to others who may be going through the same type of frustrations. If the teen is involved in LPA or another support organization, they may begin to become more interested and involved during these years. Teens who are involved in LPA are very enthusiastic about dating, and often learn how to manage long-distance relationships. They can blend their friend groups by taking their dates to dances or proms, but at times, they may be more interested in their dwarfism peer group. This is natural, and often ebbs when they go off to college, where young adults seem to have enthusiasm for both their average stature and short stature friends.

PREPARATION AFTER HIGH SCHOOL
Transitioning to College and the Workplace

Paying for College

Scholarships: There are several scholarships out there for people with disabilities. Little People of America has a robust scholarship program and receives support from the Greenberg Center at Johns Hopkins to award scholarships for both undergraduate and graduate students, as well as students who are pursuing an education from a trade school.

Vocational Rehabilitation Programs: Vocational Rehabilitation Programs are typically managed by each state's Division of Rehabiliation Services (DORS or DOVRS). Services provided by these programs assist with pre-employment transition services, including funding for college. Students must have a documented disability, and if they had a 504 or IEP plan in secondary school, this is helpful in determining eligibility. According to Federal Law, students with the most significant disabilities must be served first, so the amount of assistance provided will depend on the state's funding.

Selecting a College

Disability Support Services: Most colleges have a department or office dedicated to students with disabilities. It may be useful for the prospective student to contact the disability office to find out what kind of accomoodations the college can offer, from stools in classrooms to accessible dorms.

Visiting the Campus: Each college is unique, and it's important to visit the campus to find out how easy or difficult the landscape is, how open the students and teachers are to differences, and the general feel for the environment. Becoming involved with activities on a smaller scale on campus before the all of the students arrive can be helpful in making connections and friendships.

SPORTS PARTICIPATION

Competitive

Children with achondroplasia who are interested in sports can usually compete on a typical team with average height peers until around ten years old. If a coach is especially supportive of the child with achondroplasia, involvement with an average stature team may be possible after he/she is 10 years old. However, it is around that time when sports becomes more competitive children with dwarfism or other differences are 'benched' more often, and are less likely to receive equal playing time as their average height peers. Sometimes, if the child/teen has a true passion for the sport, they may find that being a manager of their high school team is a good way to stay involved. However, there are other outlets available to them to play the sport itself.

Recreational

Recreational sports are a wonderful way for children and teens to hone skills in a particular sport and learn valuable skills in teamwork. One of the keys to a successful experience is finding a coach who Is willing to allow the child with achondroplasia to play. Finding different outlets for recreational sports may be the best bet; there are adaptive sport organizations all over the country which are open to people with all types of physical disabilities. Not only do these organizations provide a non-judgemental place to play sports, they also allow children with achondroplasia to connect and network with children with other disabilities.

SPORTS ORGANIZATIONS

Dwarf Athletic Association of America (DAAA)

The DAAA ("D triple A") is an organization created in 1985 to provide athletic competition opportunities to athletes with dwarfism. They host their National Games in partnership with the LPA national conference every July. Participation is offered to kids and adults, from 4 years old and up. Sports offered include basketball, soccer, flag football, table tennis, boccia, badminton, swimming, track and field, and volleyball. It is a great way for short statured children to compete on a fair level.

United States Paralympics

There are several sports for elite athletes with achondroplasia within the Paralympics. Like the Olympics, there are winter and summer games for multi-sport events. Athletes with a range of physical disabilities have competed worldwide since it began in Britain in 1948. Achondroplasia is included within the Paralympic's short stature classification, which is usually referred to as S6. Paralympic short stature sports include badminton, equestrian, javelin, long jump, powerlifting, swimming, table tennis, and wheelchair tennis.

Achondroplasia Over the Lifespan: ADULTS

HIGHLIGHTS:

1. Preservation of physical function and back health: Stenosis (narrowing) in the spine causes the great pain in adults with achondroplasia. Avoid prolonged unsupported sitting, support your legs with stools or footrests while sitting and maintain a healthy weight.

2. Weight Management: Maintaining a healthy weight is essential to remain physically active. Working with a nutritionist to establish a well-balanced and calorically sound diet may be needed. Bariatric surgery is an option if excessive weight is jeopardizing activity and survival.

3. Sleep disordered breathing: If there is excessive daytime sleepiness, inability to concentrate at work, heavy snoring or observed apnea, obtain a sleep study to assess for sleep disordered breathing.

4. 'Owning' your medical care: As a teen transitions into adulthood, it is essential for the individual with achondroplasia to take over the responsibility of his/her medical care from their parents or caregivers.

5. Maintain routine medical care outside of achondroplasia: The adult with achondroplasia should be treated by their primary care physician the same as all other adults in terms of health maintenance and surveillance (e.g. check blood pressure, lipid panels, mammograms, colon cancer screening).

6. Driving: Pedal adaptors are readily available and easy to install to allow short stature adults with drive independently.

7. Audiology: Annual audiology assessment is recommended.

8. Inheritance and genetics: There is a 50% chance for a person with achondroplasia to pass the condition on to each child. This recurrence risk changes if their partner also has a short stature dysplasia. Genetic counseling is advised to discuss recurrence risks and review genetic testing options. **See pages

9. Reproduction: There is no evidence that fertility is different in women with achondroplasia than it is in average stature women. A woman with achondroplasia will require a C-section to deliver a baby due to the size of her pelvic opening. Ideally a woman with a skeletal dysplasia diagnosis who would like to pursue pregnancy would have a full neurologic exam and imaging of her spinal cord to determine the best delivery anesthesia option for her. **See pages

10. Drawing blood: Blood draws can be challenging due to decreased elbow extension in patients with achondroplasia. If unable to extend arm and visualize the antecubital veins, have the patient place the fist of their non-blood drawing arm under their elbow to change the position. You may need to use veins in the hands to obtain a blood sample.

11. Checking blood pressure: Checking blood pressure in the upper arm may not be possible. It is better to use the forearm to check blood pressure than to stop checking blood pressure. As in all adults, it is important to make repeated measurements of blood pressure before embarking on treatment of hypertension.

12. Know your rights when it comes to access to your world. Check the Americans with Disabilities Act website for access to important information and resources. www.ada.gov

Psychological Health

Talking with friends and family about achondroplasia

Both parents of children with achondroplasia and children themselves will be asked about their stature by friends, family, neighbors and strangers. Sharing information with friends, family, teachers and coaches is often desired to allow important people in the child's life to understand their diagnosis and any necessary accommodations. Sharing factual information in a straightforward manner is often the most effective. The manner in which parents/caregivers address questions about the child with achondroplasia is the example to the child and his/her siblings about how to handle such situations. Many questions to children from peers are often due to curiosity about the child's age and can be addressed directly and simply.

The "Elevator Speech"

For the child with achondroplasia, he or she should have a 30 second or less practiced response to address questions about their stature/diagnosis by about 5 years old. Having a response that the child's family has time to develop and practice provides the child confidence and empowerment to answer questions and develop autonomy. It is similarly important to educate children that they always have the option to not answer questions about their stature, particularly when it is from strangers.

Bullying

Bullying can occur to anyone, particularly those with physical or mental differences. Short stature is visible to everyone so children and even adults with achondroplasia (or other short stature dysplasias) may experience bullying. It is important to educate the child that bullying is not acceptable and will not be tolerated. The child should know how to tell an adult if he/she is being bullied at school, home, in activities. In many instances, if the bullying occurs at school, the administration can discipline the offenders without letting them know that the child had alerted them to the incidences. ("Another student saw you push Jon/Suzy and was concerned…")

Patient support groups, social support

- Little People of America
- Little People UK (LP UK)
- Little People Ontario
- Short statured People of Australia (SSPA)
- Understanding Dwarfism

Siblings

Sibling rivalry can happen in any family, but at times the sibling relationship can be challenging when one of the siblings has achondroplasia.
From time to time, average stature siblings may feel:

- Responsible for brother/sister and feel stressed out when there is teasing or unfairness.
- Neglected by their parents because of the time spent on medical appointments or other activities.
- Resentment toward sister/brother on how much outside attention they receive.
- Guilt /mixed emotions about resenting their brother/sister.

LPA hosts a sibling group at the national conferences which allows siblings to talk confidentially about these feelings, and provides networking for support during the year so that the child knows they are not alone in feeling a certain way.

Disability coverage decisions

The diagnosis of achondroplasia may be considered a disability, depending on how the condition limits a child's activities. There are also income eligibility limits that would be considered in an application. There is more information in the Benefits for Children with Disabilities booklet published by the Social Security Administration. There is some controversy regarding applying for Supplemental Security Income (SSI) or Social Security Disability Insurance (SSDI) within the dwarfism community. Some dwarfism advocates feel achondroplasia and other dwarfism is not a disability at all, and the environment, including attitudes, needs to be more accepting of differences. This is you and your family's decision about how to consider the diagnosis of achondroplasia.

Common medical treatments for people with short stature

Audiology: This includes tests of hearing, balance and other ear function. A person must hear to develop speech and learn from their environment. An audiologist is the person who performs these tests. He/she works with an otolaryngologist (ear-nose-throat, ENT, doctor) to determine if the patient needs surgery to treat chronic ear fluid or airway obstruction or hearing aids to improve hearing. A child with achondroplasia should have audiology testing and ENT assessment every 6-12 months and annually as an adult.

Sleep Study: For children, a sleep study should occur overnight in a sleep laboratory in an age-appropriate bed where a parent can stay overnight. Ideally this occurs in a pediatric sleep laboratory where a skilled technician places the sticky leads on young patients' head to measure breathing rate and oxygen levels. In adulthood, studies may occur at home under the direction of a pulmonologist (lung doctor). Sleep studies assess for obstructive sleep apnea and central sleep apnea, both common problems for people with achondroplasia.

MRIs: Magnetic resonance imaging is a radiology study used in people with achondroplasia to examine the brain and spinal cord. There is no radiation exposure with an MRI. In young patients, sedation may be needed to prevent movement and obtain clear images. Sedation/anesthesia should only be administered in a setting where a physician or nurse anesthetist skilled in difficult airway management is present.

X-rays: X-rays of the back and legs are performed to assess for kyphosis (i.e. posterior curvature of the mid to lower spine) and leg malalignment/bowing. The orthopedist assesses these films in conjunction with their physical examination to determine when or if surgery or other intervention is needed.

Orthodontia/dentistry: All children, including those with achondroplasia, should have regular dental assessments and cleaning every 6 months as soon as teeth erupt. Dental crowding and tooth malalignment are common in people with achondroplasia due to relatively smaller space for teeth. Adding an orthodontist to the healthcare team is recommended.

Common surgical treatments for people with achondroplasia

Tonsillectomy and/or adenoidectomy: This is the surgical removal of the tonsils and/or adenoid (i.e. pads of immune-related tissue) from the back of the throat. In people with achondroplasia, this procedure is performed because the tonsils/adenoids may block the air passage and cause obstructive sleep apnea (OSA). OSA is treated surgically if a sleep study indicates the person's oxygen level while sleeping is too low or the carbon dioxide level is too high due to pauses or difficulty breathing. Sometimes the adenoids are removed first and then the tonsils or both are surgically removed together; this depends on the age of the child and the severity of the obstruction. An otolaryngologist (ENT surgeon) performs this procedure.

"Tubes": This term refers to the surgical placement of an extremely small appliance ("tube") into one or both ear drums of a person with achondroplasia to drain fluid that accumulates behind the ear drum. Other terms for this appliance are pressure equalizing tubes, tympanostomy tubes or grommets. Drainage of chronic middle ear fluid is important to promote better hearing, develop speech and prevent infections. An otolaryngologist performs this procedure.

Cervicomedullary decompression (CMD): This is a surgical procedure performed by a neurosurgeon to relieve compression by the bone of the skull and the vertebrae on the upper spinal cord and brainstem. MRIs are utilized to detect compression which necessitates the surgery and to assess the result. This procedure is done when compression of the spinal cord/brain stem is causing dangerous pauses in breathing, weakness, pain, poor feeding or choking.

Spinal decompression and fusion: This is a surgical procedure performed by a neurosurgeon and/or orthopedic surgeon to relieve compression by the vertebrae on the spinal cord. This procedure is carried out when compression of the spinal cord is causing severe pain, weakness and/or decreased endurance. Sometimes the spinal compression causes numbness or tingling in the extremities; this is not normal and should be discussed with your physician, if present.

Lower extremity realignment: In patients with achondroplasia, the ends of the bones around the knee are irregular and wide which cause the knees to curve outward (bow-legged). This can cause pain, an irregular gait and decreased physical activity. An orthopedist can cut the bones and realign them with surgical pins, plates and/or casts.

New drug therapies for achondroplasia

The information on this page represents the state of pharmaceutical development for young patients with achondroplasia, as of 2022. The authors of this brochure are not representing or promoting specific companies developing these research medications for people with achondroplasia. The purpose of this content is to inform patients, their families and care providers about these research medications and provide resources to learn more, if desired.

Recent Developments

Several pharmaceutical companies have developed medications to treat short stature in young patients with achondroplasia. These medications area being studied in children with achondroplasia in clinical trials. The purpose of these clinical trials is to determine if the medications are safe for use in people and if they affect bone growth.

Clinical trials are required by the Food and Drug Administration (FDA) to test new medications before they are approved and become available by prescription. At this time, one medication is available by prescription (i.e. Voxzogo). The other research medications are currently available only by participating in these clinical trials. Some of the research medications for patients with achondroplasia area given by mouth while others are given in the form of an injection (i.e. a shot).

All clinical trials occurring in the US must be listed on the US government website, clincialtrials.gov. Some of the clinical trials for infants and young children with achondroplasia listed on this website also include information about research occurring outside the US. The website includes contact information and internet links to the companies. Check clinicaltrials.gov for more information about these and future trials with patients with achondroplasia.

Resources

- **Clinicaltrials.gov** An online resource managed by the National Library of Medicine at the National Institutes of Health
- **who.int/health-topics/clinical-trials#tab=tab_1** An online resource about clinical trials managed by the World Health Organization

Differences in medical & surgical treatment around the world

Limb Lengthening: Perspectives on limb lengthening vary in dwarfism support groups and clinics around the world. In the US, the Little People of America (LPA) does not promote limb lengthening, but accepts members who have had the procedure. In Spain, the Fundacion ALPE Achondroplasia (ALPE) supports treatments for achondroplasia, including limb lengthening. Italy's patient support organization, the AISAC, supports medical intervention and limb lengthening on their website. The majority of children in Spain and Italy with achondroplasia begin limb lengthening at a young age. People in Japan with achondroplasia often have limb lengthening in combination with growth hormone treatment.

Limb lengthening may be performed on the legs and arms. As with any surgical procedure, there are potential complications. If a young patient with achondroplasia (and their family) opts for limb lengthening, an experienced surgeon with a team to determine the suitability of that child to have this prolonged procedure and manage

its short- and long-term complications are helpful.

Growth Hormone Therapy: Growth hormone (GH) has been used to treat short stature in achondroplasia. It is widely used in Japan but achondroplasia is not an indication for treatment in the US. A recent meta-analysis of 12 studies of nearly 600 children with achondroplasia treated with growth hormone showed there was an increase in growth in the first year that was not sustained over a longer period of time (see references). Having achondroplasia does NOT mean the person is also growth hormone deficient—these are two separate medical conditions.

MRIs and sleep studies: In some infants and young children with achondroplasia, the foramen magnum (hole at the base of the skull next to the upper neck) is too small and the spinal cord is compressed. This can cause severe neurologic problems (even death) if unrecognized and not treated. To assess for compression, all infants and young children with achondroplasia should have regular physical examinations with a neurologic exam, assessment of development and growth, and ideally an MRI of the neck and brain and a sleep study. In some medical settings, MRIs and sleep studies are not readily available. If there are abnormalities in the neurologic exam, in growth or development, every effort should be made to consult with a neurosurgeon as soon as possible and obtain the MRI and sleep study.

Genetics of achondroplasia

Achondroplasia is a dominant genetic condition caused by a difference in the DNA sequence of the FGFR3 gene, called a variant (previously termed 'mutation'). Every person with achondroplasia has a variant at the 1138th letter of DNA in one copy of their FGFR3 genes. Instead of a 'G' for guanine, there is an 'A' for adenine or a 'C' for cytosine. This is represented by c.1138G>A or c.1138G>C on the genetic test report. This letter replacement causes the FGFR3 gene to make an abnormal protein that hinders growth of the long bones of the limbs, the digits and the base of the skull. This causes short stature and the other features of achondroplasia.

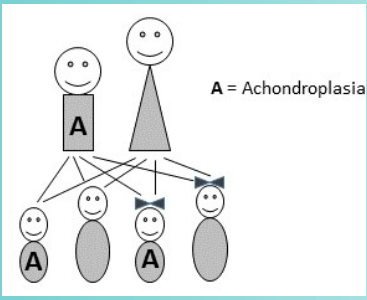
People cannot be average stature 'carriers' of achondroplasia. Every person with the 1138 FGFR3 gene variant will have short stature and the other features of achondroplasia.

The FGFR3 gene variant that causes achondroplasia can be inherited from a parent who also has achondroplasia, or, it can occur spontaneously as a new DNA variant. The majority (80%) of people with achondroplasia have a new (not inherited) DNA variant; their parents have average stature.

These spontaneous DNA FGFR3 gene variants occur in about 1 in 20,000 to 30,000 babies. These variants occur through no fault of either parent or due to the use of any medication or other exposure prior to or during the pregnancy.

For average stature parents who have had a child with achondroplasia, there is a 1-2% chance in each future pregnancy to also have achondroplasia due to gonadal mosaicism. This is one or more remaining eggs or cells which make sperm having the same spontaneous variant even though the person does not have achondroplasia.

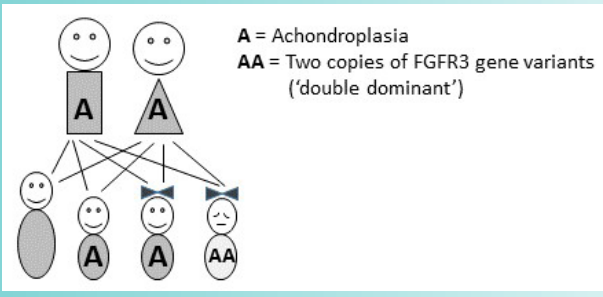
Males and females can pass on achondroplasia.



Autosomal dominant: With every pregnancy, 50% chance of achondroplasia and 50% chance of average stature.

For a person with achondroplasia, there is a 50% (1 in 2) chance in each pregnancy he or she will pass on the FGFR3 gene copy with the 1138 variant and have a child with achondroplasia. There is also a 50% (1 in 2) chance in each pregnancy to pass on the other FGFR3 gene copy and have a child with average stature.

Both parents may have achondroplasia.

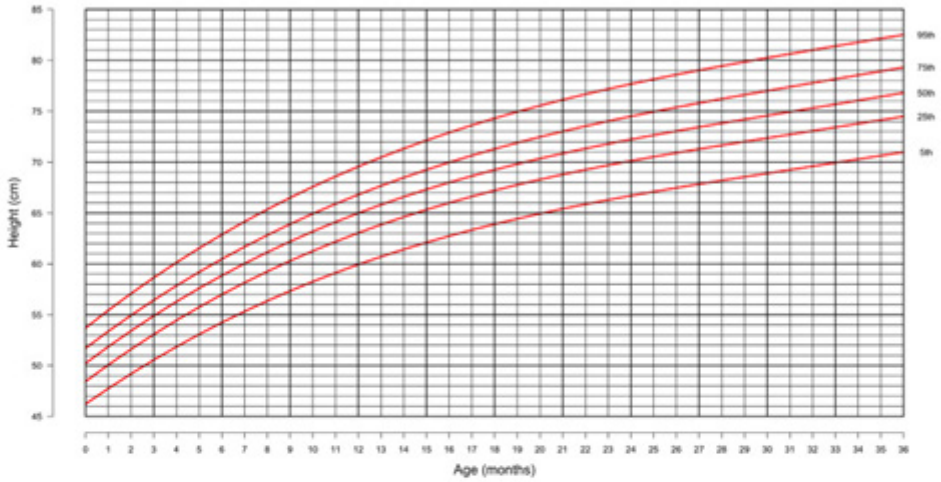


Double dominant: An infant born with two 1138 FGFR3 gene variants cannot survive due to small chest and lungs.

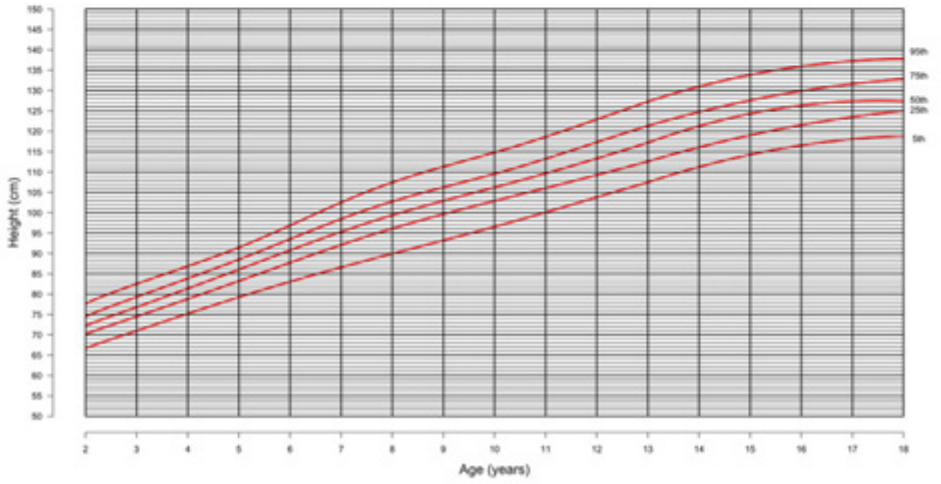
Two parents with achondroplasia could have a child with achondroplasia (50% chance), average stature (25% chance) or 'double dominant' (25% chance) meaning the child inherited both parental 1138 FGFR3 gene variants. Infants with 'double dominant' achondroplasia might survive short-term but only with heroic, lifelong medical intervention.

RESOURCES - GROWTH CURVES

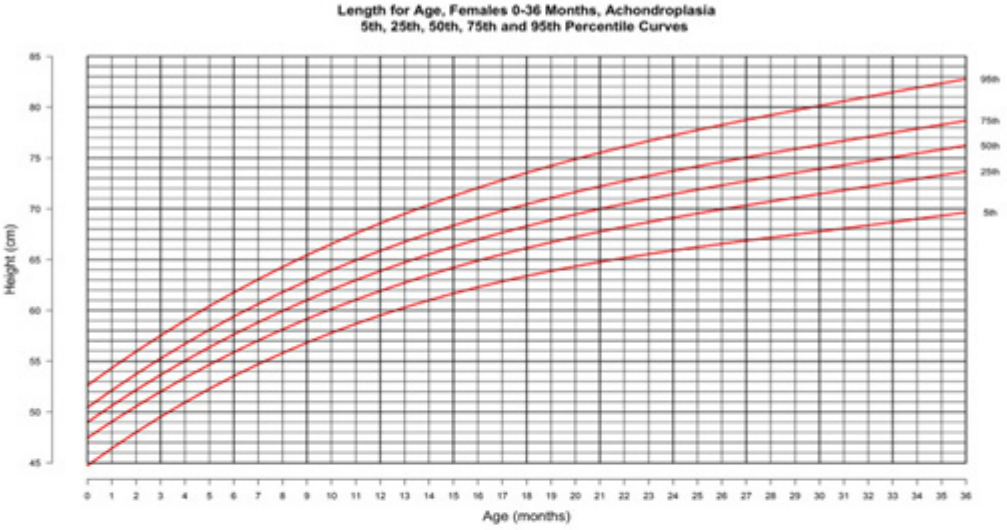
Length for Age, Males 0-36 Months, Achondroplasia
5th, 25th, 50th, 75th, and 95th Percentile Curves

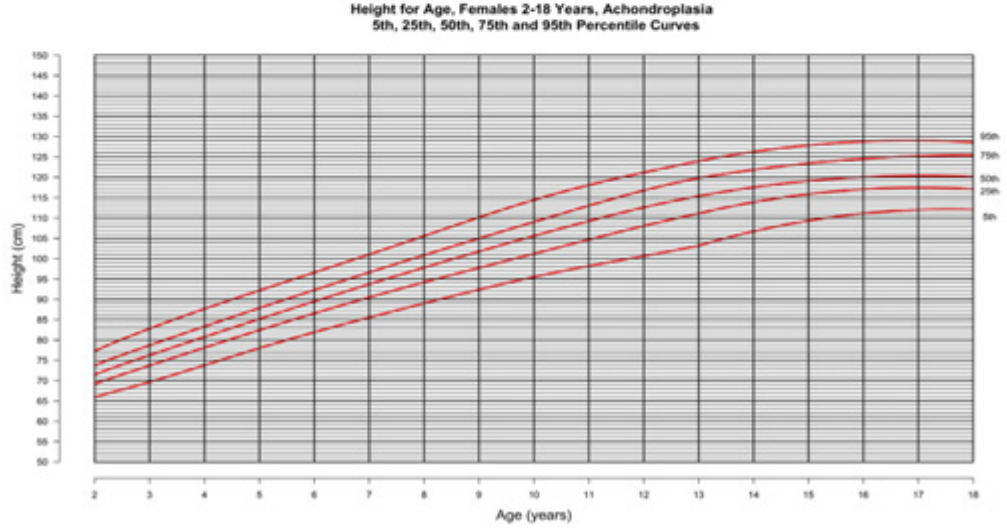


Height for Age, Males 2-18 Years, Achondroplasia
5th, 25th, 50th, 75th and 95th Percentile Curves

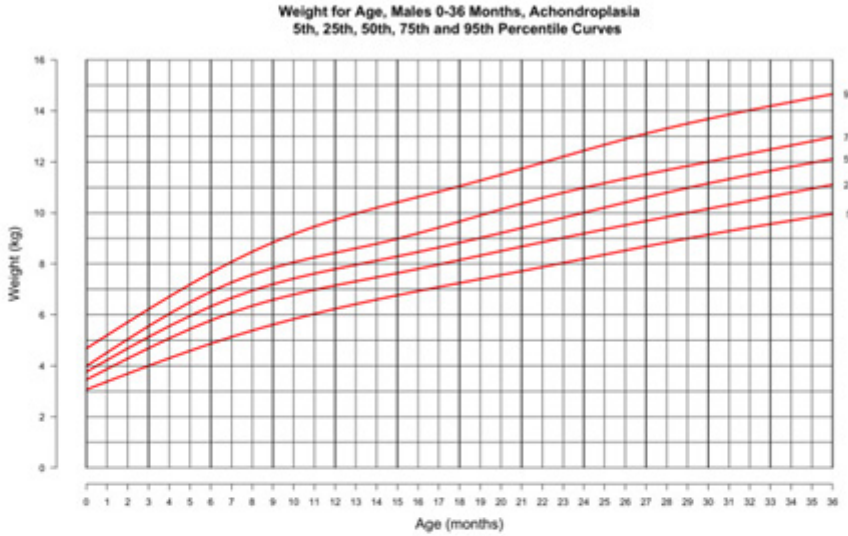


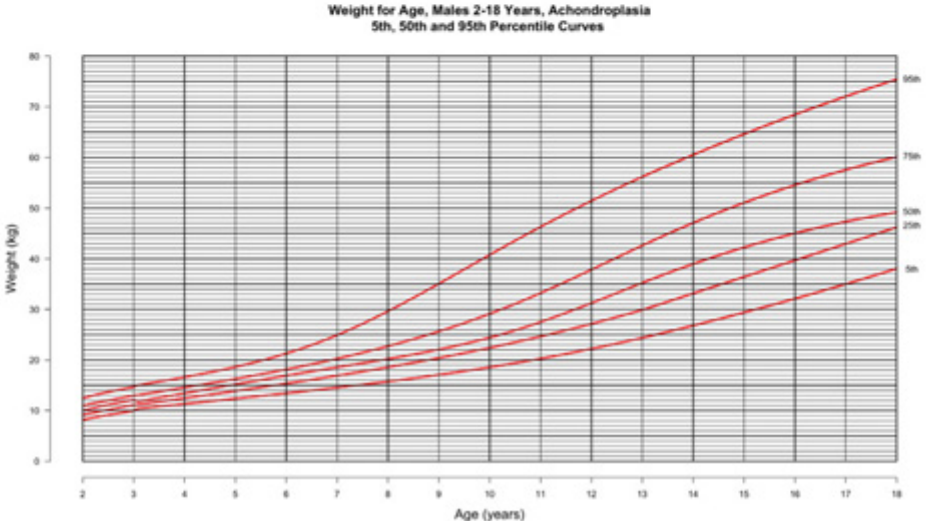
Length and Height for Age



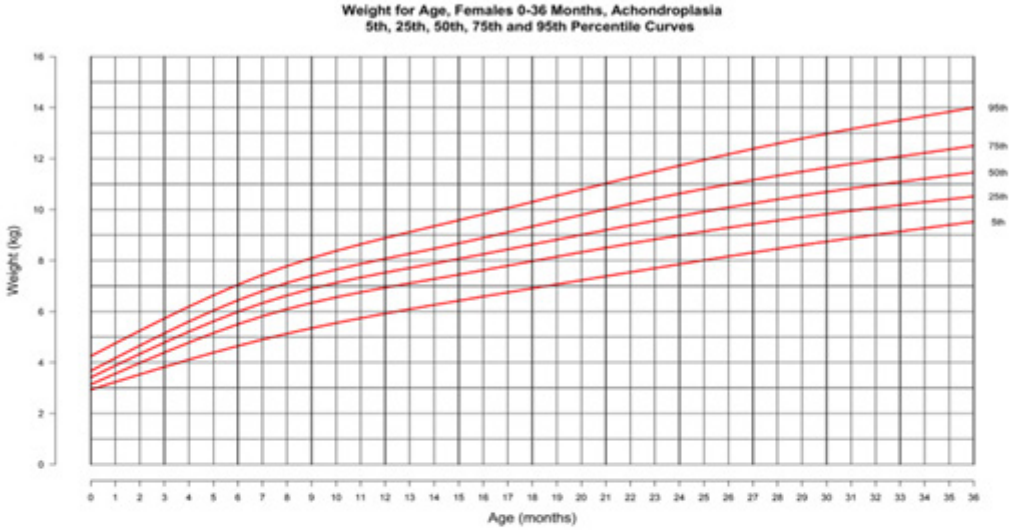


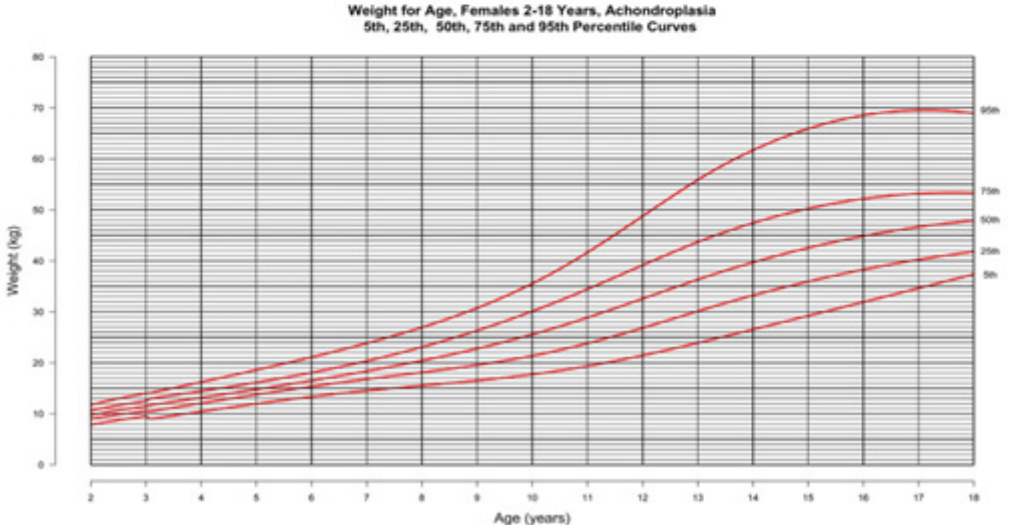
RESOURCES - GROWTH CURVES





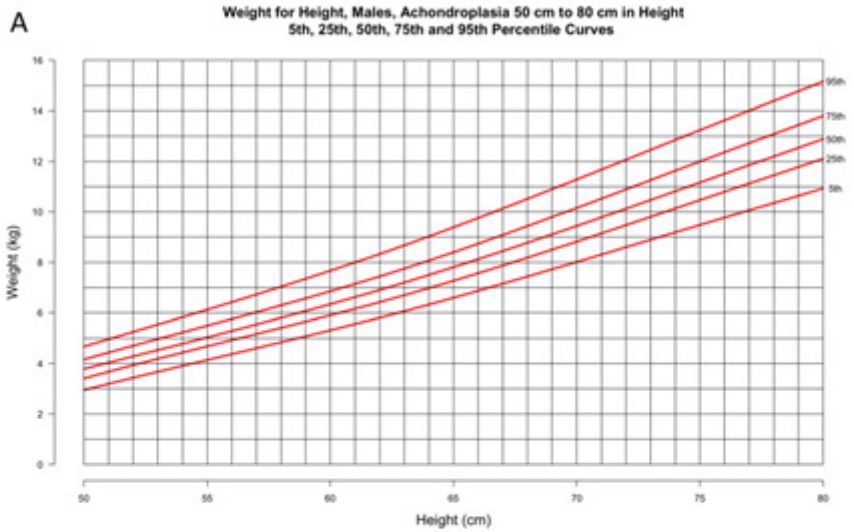
Weight for Age





RESOURCES - GROWTH CURVES

A



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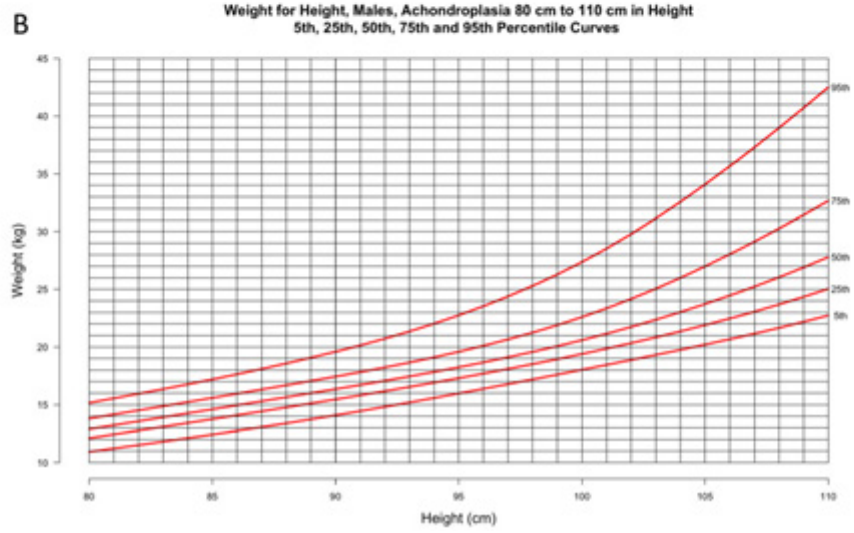
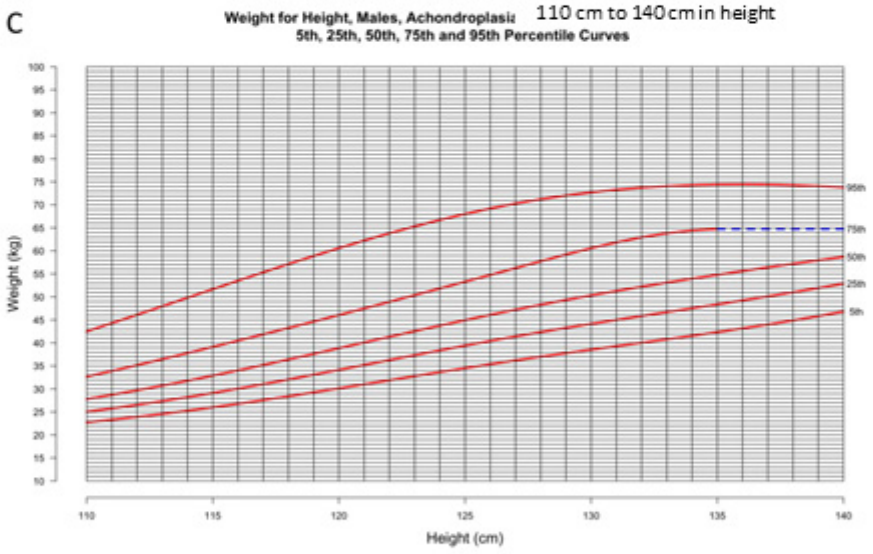


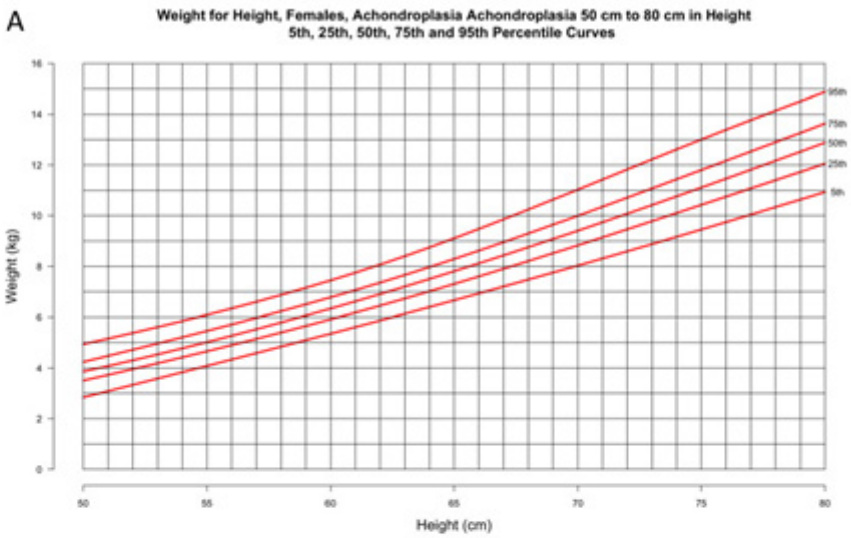
Weight for Height, Male

C



RESOURCES - GROWTH CURVES

A



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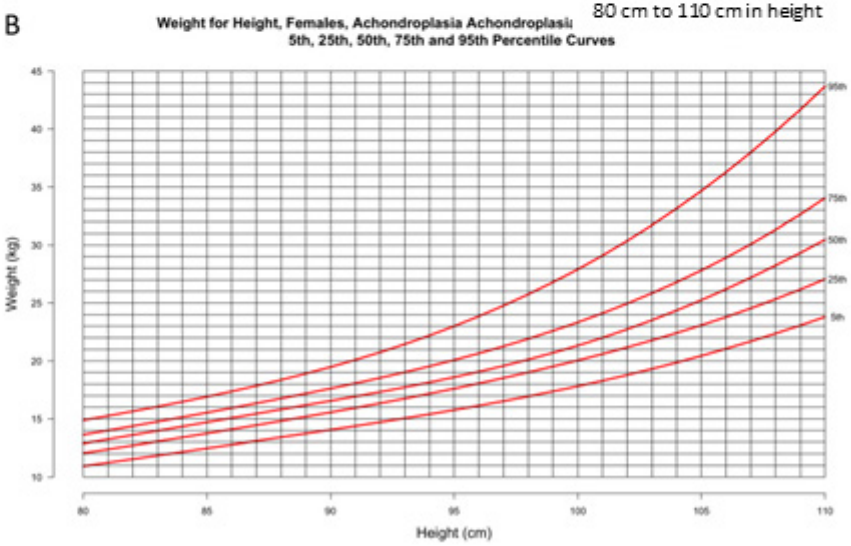
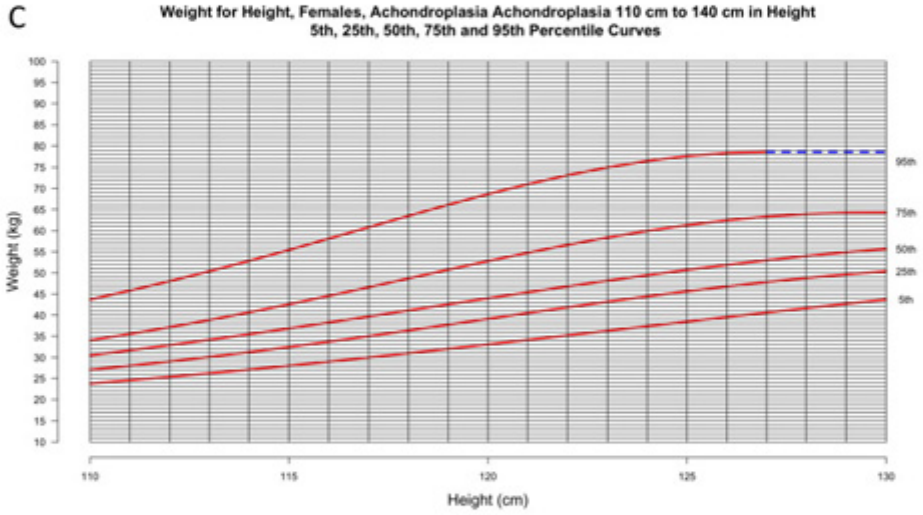
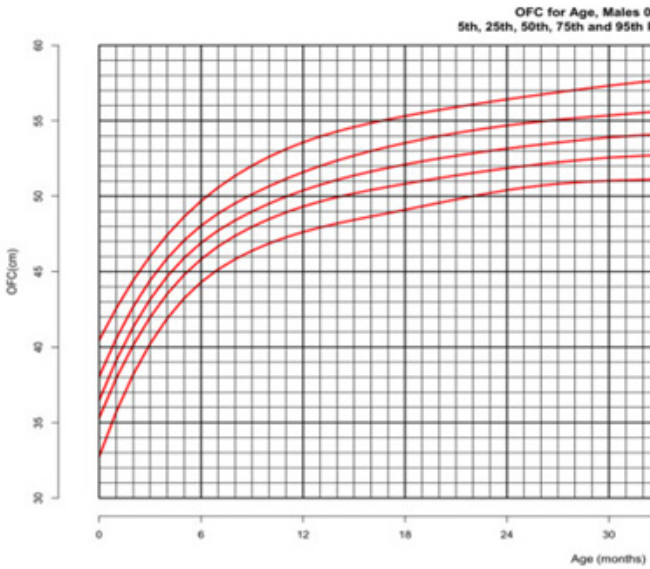


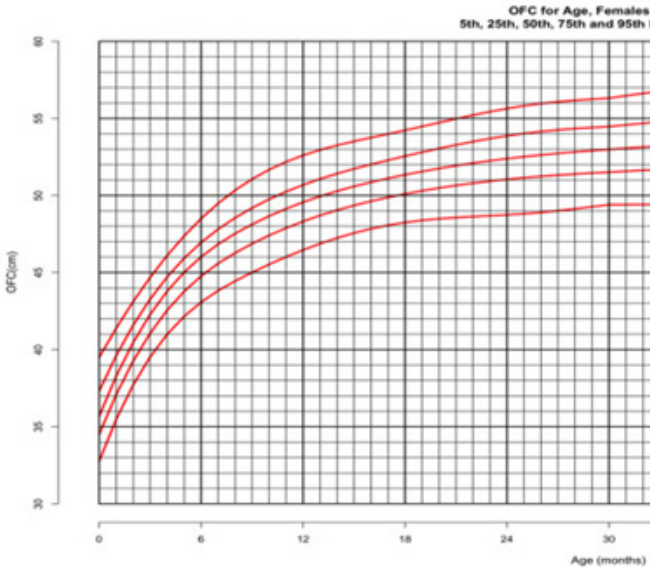
Weight for Height, Female

C



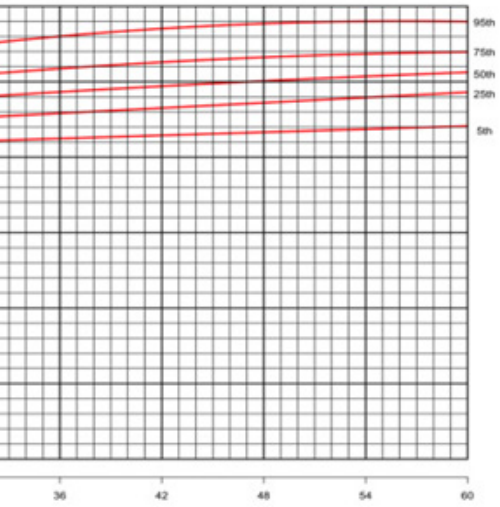
RESOURCES - GROWTH CURVES



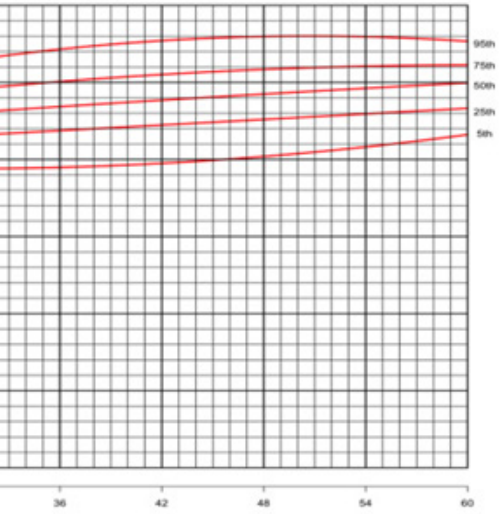


Head Circumference

-5 Years
Percentile Curves



0-5 Years
Percentile Curves



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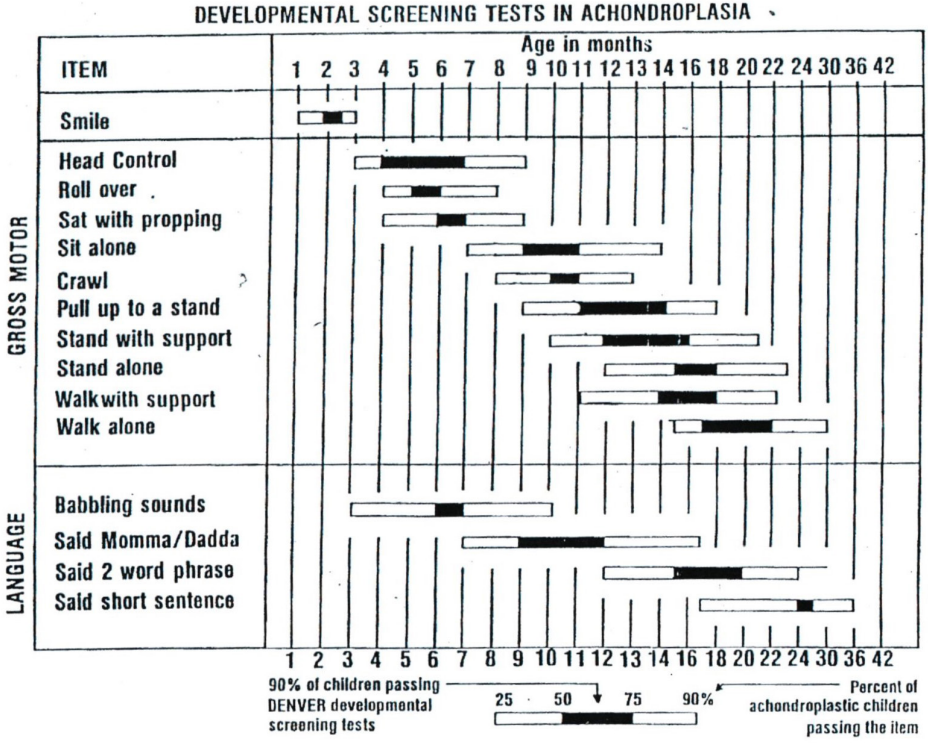
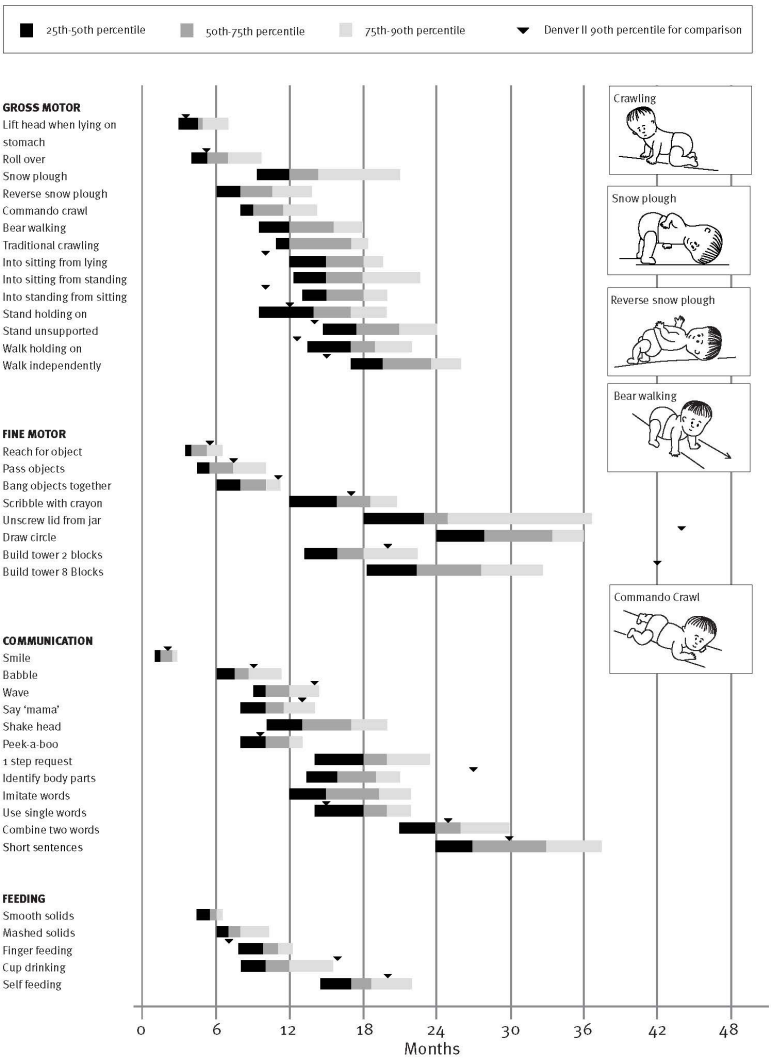
Developmental Milestones for infants and young children with achondroplasia



Achondroplasia Developmental Recording Form

Name:	Examiner's name:
Date of assessment:	Age of child (yrs/months):



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RESOURCES - Helpful Links

Clinical Information:

- Achondroplasia - GeneReviews® - NCBI Bookshelf (nih.gov) (pg 3)
- Achondroplasia: MedlinePlus Genetics
- Autosomal Dominant Inheritance: fact-sheet-8-autosomal-dominant-inheritance (genetics.edu.au)

Car Seats:

- NHTSA Car Seat Information: Car Seats and Booster Seats | NHTSA (page 10)
- SafeRide4Kids State Car Seat Laws: Car Seat Laws by State - Find your state car seat laws (saferide4kids.com) (page 10)

Education & Disability Resources:

- 504 Plans and IEPs: 504 Education Plans (for Parents) - Nemours KidsHealth (pages 12-16)
- Parent and Educator Resource Guide to Section 504 in Public Elementary and Secondary Schools (pages 12-16)
- IEP Resources through Parent Center Hub: All About the IEP | Center for Parent Information and Resources (parentcenterhub.org) (page 16)
- Benefits For Children With Disabilities (ssa.gov) (page 19)
- Adult Transition & Employment Resources: Division of Rehabilitation Services (DORS) | Maryland PROMISE (mdtransitions.org) (Maryland-specific, refer to your state's Vocational Rehabilitation or Social Services Department Website)
- Your Legal Disability Rights | USAGov
- ADA.gov homepage (Americans with Disabilities Act website)

Dwarfism Awareness:

- Dwarfism Awareness Month (lpaonline.org)
- Rare Disease Day | Nation Organization for Rare Disorders (rarediseases.org)

Books (LPA):

- Fiction with Dwarf Characters (lpaonline.org)
- Children's Dwarfism Book - I'm Just Small (imjustsmall.com)
- Parenting and Childrens Books (lpaonline.org)
- Children's Book — Understanding Dwarfism "Not Too Big... Not Too Small... Just Right for Me!

Support Groups:

- Little People of America
- Little People UK
- Little People of Ontario
- SSPA - Short Statured People of Australia
- Understanding Dwarfism

Miscellaneous:

- Stop Bullying
- Adaptive Products
- Positioning and Handling babies with achondroplasia

RESOURCES - NOTES

Clinical Trials, Research, Therapies:

- Clinical Trial Information: Home - ClinicalTrials.gov
- World Health Organization Clinical Trial Information: Clinical trials (who.int)
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