

Achondroplasia at a Glance

Achondroplasia is a genetic condition caused by a change in a gene that provides instructions for making a protein used to form bone and brain tissue. Achondroplasia is the most common form of inherited disproportionate short stature, or dwarfism. The word achondroplasia means “without cartilage formation.”

Individuals with achondroplasia don’t convert cartilage into bone, particularly in the long bones. Achondroplasia is inherited.

However, about 75% of people with achondroplasia have a new mutation and have average size parents.



Meet Lacey-Mae on page 8

About 1 in every 26,000-28,000 babies is born with achondroplasia.

Typically, individuals with achondroplasia have normal intelligence and a normal life span.

General Dwarfism information:

There are over 200 distinct types of dwarfism (generally a term used for a person 4’10’ or under). About 1 in every 10,000 babies is born with dwarfism. The most common dwarfing condition is achondroplasia.

However, there are many different reasons a person may be shorter than average. The most common reasons for short stature in children are familial short stature, a difference in the timing of growth (called constitutional growth delay) and, in girls, Turner syndrome. In addition, a person may be short for many other reasons (i.e. enzyme processing function disorder, kidney disease, etc.)

In general preferred terms are short stature, little person (LP), dwarfism, and for some people dwarf. *However, as with all medical conditions, the preferred terminology is the **person’s name**.*

Things to Think About

1. Medical/Dietary Needs

What you need to know

The list of *possible* medical problems in achondroplasia can be extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely between individuals. Therefore, it is important to ask the parents about the medical issues in their child.

- School age children with achondroplasia may have annual doctor and specialist visits to monitor medical conditions.
 - Routine management of middle ear infections and monitoring of hearing should be undertaken
 - Obesity should be avoided to reduce risks associated with airway obstruction and musculoskeletal limitations
 - Bracing may be needed for severe kyphosis and knee instability
- Be aware, or ask parents, if the child has a medical alert bracelet.
- No special diet is required for achondroplasia, although a well-balanced diet is important.

Physical characteristics and/or symptoms:

Not all people with Achondroplasia have all of these characteristics.

- **Short stature** caused by shortening of the limbs
 - Average height: men: 131cm
 - Average height women: 124cm
- **Intelligence** is usually typical
 - Unless hydrocephalus or other central nervous system complication is present.
- **Obesity:**
 - In adults this can:

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- Contribute to morbidity associated with lumbar stenosis
 - Contribute to joint problems
 - Lead to possible cardiovascular complications
- **Spinal curvature:**
 - Lumbar lordosis where the spine curves inward at the lower back.
 - Kyphosis characterized by an abnormally rounded upper back
- **Joint issues:**
 - Limitation of elbow extension and rotation
 - Excessive mobility of the knees, hips, and most other joints

Other findings:

- Most common adult medical complaint is symptomatic spinal stenosis involving L1-L4
 - Symptoms range from intermittent, reversible, exercise-induced limping to severe irreversible abnormalities of leg function.
 - Spinal stenosis- may manifest in older individuals with numbness, weakness, and altered deep tendon reflexes
- Sleep apnea
- Middle ear dysfunction
 - If inadequately treated this can lead to hearing loss that may interfere with language development
- Bowing of legs
- Characteristic facial features

What you can do

- A yearly check up and studies as needed should occur in the child's Medical Home.
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.

2. Education Supports

It is important to have HIGH LEARNING EXPECTATIONS for children with Achondroplasia. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

What you need to know

Individuals with achondroplasia typically have normal intelligence. A small percentage of individuals may have intellectual delay due to complications of hydrocephalus or for a reason unrelated to achondroplasia.

It is important to find the balance between providing help and fostering autonomy. Individuals with achondroplasia are able to live independent productive lives with adaptive, adjustments, or assistance. An IEP/504 may be in place for individuals' safety and comfort in the class and school.

What you can do

- Adaptive aids in school may be required for
 - Heavy doors
 - High doorknobs
 - Reaching the blackboard
 - Extenders
 - Stools
 - Desk size
 - Bathroom
 - Use regular bathroom with a permanent step
- Carrying books may be challenge
 - Two sets of books; one for home one for school
 - Friend helper
 - Low locker
- Allow extra time to travel between classes/use elevators
- May need stool to rest legs on
 - Legs may fall asleep if left to dangle
 - Upper legs are too short to allow back support. Use a pillow for back support.

- Speech therapy may be necessary, especially if concerns with hearing loss
- Occupational therapy and/or accommodations for writing
 - Individuals may have small fingers and joint hypermobility due to stiffness
 - May not be able to write at a quick enough speed
 - Consider tape recorder for class
 - Use computers
 - Additional time for tests or provide oral exams

3. Behavioral and Sensory Support

What you need to know and what you can do

- It is important to treat an individual age appropriately
 - Treat people respectfully and not as a young child just because they are small.
- Treat them with dignity and do not carry people who can walk. Individuals may feel social stigma due to short stature
 - Little People Association has many resources
 - Counseling may be appropriate
 - Support groups

4. Physical Activity, Trips, Events

What you need to know

- Exercise and physical education should be encouraged for strength building and obesity prevention
 - PE programs can be modified
 - Goal is for child to finish at same time as other children (i.e. run 2 laps instead of 4)
 - Downsize equipment – i.e. smaller bat
 - Have a designated runner
 - Keep them safe and involved.

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- Choose activities to include children as appropriate if activity can be modified.
 - Swimming and biking are encouraged
- Care must be taken to limit risks for injury to the spinal cord at the craniocervical junction. They have decreased size of spinal column surrounding the spinal cord in the neck area. It is important to be aware of head and neck trauma.
 - This includes avoiding:
 - Collision /contact sports
 - Football
 - Hockey
 - Rugby
 - Use of trampoline
 - Diving from diving boards
 - Gymnastics
 - Hanging from knees or feet on playground equipment

What you can do

- Occupational therapy evaluation may help with accommodations and modifications
- Field trips
 - If a lot of walking is necessary, be aware it will take more time and individuals may be tired
 - Consider cutting down on walking when possible
 - Use alternative forms of transportation
- Assistive device are available to improve accessibility and independence in homes, school, automobile and workplace

5. School Absences and Fatigue:

What you need to know

- Students shouldn't have excess school absences
- Apnea may lead to day time sleepiness

6. Emergency Planning:

What you need to know

- There should be a plan for a emergency evacuation
 - Need to avoid situations where individuals may be trampled
 - Need to be able to reach doors

What you can do

- Assign someone to help individuals in an emergency, and identify a back-up person

7. Resources:

GeneReviews

Gene Reviews – *learn more about the genetics of achondroplasia*
<http://www.ncbi.nlm.nih.gov/books/NBK1152/>

Little People of America

<http://www.lpaonline.org/>

Little People of America (LPA) is dedicated to improving the quality of life for people with dwarfism throughout their lives while celebrating with great pride Little People's contribution to social diversity. LPA strives to bring solutions and global awareness to the prominent issues affecting individuals of short stature and their families.

<http://www.lpaonline.org/for-parents-and-teachers>

It's a Whole New View: A Guide for Raising a Child with Dwarfism" a publication of Little People of America, Inc.

American Academy of Pediatrics: Health supervision for people with achondroplasia

<https://publications.aap.org/pediatrics/article/145/6/e20201010/76908/Health-Supervision-for-People-With-Achondroplasia>

This AAP report is designed to help the pediatrician care for children with achondroplasia and their families.

Medline

<https://medlineplus.gov/genetics/condition/achondroplasia/>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

Meet a Child with Achondroplasia – Lacey-Mae, Girl with a Cause



Lacey-Mae bedazzles with her chatter and charm. From her early experience in Toddler pageants, she has gained confidence in herself and in social situations. Kerry Ann, her mother says Lacey-Mae is a ‘social butterfly’ and that she is bubbly, very talkative and outgoing. At home with 2 siblings and 2 foster children, she is one of the older children.

Lacey-Mae was born with achondroplasia. She had “1/2 of her tongue” and her vocal cords were paralyzed, says Kerry Ann, which prevented sucking and she didn’t make sounds her first year, not even a cry. She was in the NICU for one month and they think that a cranio-facial nerve was paralyzed and this eventually resolved at about 1 year of age. This is not a usual finding with achondroplasia, just something that was unique with Lacey-Mae.

Lacey-Mae received Early Intervention services and attended a regular preschool in her hometown. She had a 504 plan that helped with such supports as having a water bottle with her at all times (she can overheat easily) or having some adaptive PE to prep her for regular gym classes. Although she has to be careful with her neck and jumping, Kerry Ann did not want over-precautions to limit Lacey-Mae. She has attended school with her peers all the way through!

Now 13 years old and in high school, Lacey-Mae is involved in an Anime Club and was a Girl Scout. She is no longer involved in pageantry but is finding other interests. Lacey-Mae has not had many medical issues after her early start. She had a lumbar curve in her spine early on but that is gone. An MRI of her neck shows that it is “perfect.” Her legs have a very slight amount of bowing.

Her experiences in school have generally been very good. Some teachers have patronized her or pointed out her size in an unkind way. A terrifying incident in middle school happened when she was using the steep staircase in a “mad rush” of people and she fell and was hurt in the crowd. It was written into the IEP that she should use the elevator instead. Her teachers and classmates have been very kind and accepting for the most part.

Because Lacey-Mae can’t write as fast or run as fast and typing is hard to do quickly, problems are addressed by her team.

Lacey-Mae has been an activist in an anti-bullying campaign she created called Peace by Piece which can be found on a YouTube channel and Facebook. This started after a bullying incident that led to her needing stitches. However, she and the boy who did this to her are now friends. Lacey-Mae also likes to draw and sketch (some of her sketches are pictured) and she is teaching herself to play the piano. She still loves to dance, and sings at church. She will be attending her first church mission trip this summer 2017! Lacey-Mae likes to be with people of average size, says Kerry Ann. She certainly seems to be a person who uses her voice strongly to advocate for herself and make a difference for others through her anti-bullying campaign!

Kerry Ann's Advice to Parents:

- Don't worry! If you are so worried about acceptance and the "what if's," you will prevent your child from getting anywhere.
- Treat your child like everybody else. Kids will be kids and are full of personality.
- Meet with the teachers ahead of time.

Kerry Ann's Advice to Teachers:

- Have patience. Students may have a hard time writing and running, may even get frustrated and cry. Their hands might hurt from writing. Treat them the same but allow them extra time or supports to prevent frustration. Don't let it get to that point.
- Accept that their output may need to be different - shorter, for example, such as writing one paragraph instead of 4.
- Meet with parents ahead of time. Parents may do a little explanation or presentation to explain achondroplasia to get questions out of the way. Allow questions and answers with the students, staff and parents.
- Don't be afraid! Sometimes fear of neck injury makes teachers overly concerned and cautious. Swings, slides and ladders were just fine for Lacey-Mae!