Genetic Education Materials for School Success

# Achondroplasia

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### Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.


### Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

# Achondroplasia

## At a Glance

Achondroplasia is a genetic condition caused by a change in a gene for making a protein used to form bone. 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 can be inherited, but about 75% of people with achondroplasia will have average-size parents. Typically, individuals with achondroplasia have normal intelligence and a normal life span.

Meet Lacey-Mae on page 8

## Common Features of Achondroplasia:

- Shortened arms and legs, with the upper arms and thighs more affected than the forearms and lower legs
- Large head size with a prominent forehead and a flattened nasal bridge
- Curved lower spine which may cause a small hump near the shoulders that usually goes away after the child begins walking
- Narrow spinal canal that may lead to spinal cord compression in adolescence
- Bowed lower legs
- Poor muscle tone and loose joints
- Sleep apnea
- Frequent middle ear infections that may lead to hearing loss
- Delayed developmental milestones
    - walking may occur between 18-24 months of age instead of around 12 months

*In general, accepted 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.*

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# In Depth

## Medical and Dietary Considerations

### 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
    - Spinal curvature may require bracing to manage lumbar lordosis where the spine curves inward at the lower back.
- There may be excessive mobility of the knees, hips, and most other joints.
- Elbow extension and rotation may be limited.
- No special diet is required for achondroplasia, although a well-balanced diet is important

### What you can do

- Encourage a comprehensive medical evaluation for the child each year. Get copies of any changes to medications or care recommendations.
- 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

## Education Supports

It is important to have high learning expectations for children with Achondroplasia, utilizing their strengths and interests as a foundation**.** Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

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## What you need to know

Individuals with achondroplasia typically have normal intelligence. A small percentage of individuals may have intellectual delay.

It is important to find the balance between providing help and fostering autonomy. Individuals with achondroplasia can live independent 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 and chair
    - 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.
- Bathroom - Use regular bathroom with a permanent step

Other considerations

- Allow extra time to travel between classes/use elevators
- Carrying books may be challenging
    - Two sets of books; one for home one for school
    - Friend helper
    - Low locker

Speech therapy may be necessary if there are issues with hearing loss

Occupational therapy and/or accommodations for writing

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- 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
    - Modified notetaking (i.e. fill in the blank notes)
    - Use computers
    - Additional time for tests or provide oral exams

# Behavioral and Sensory Support

It is important to treat an individual age appropriately. Treat people respectfully and not as a younger child just because they are small.

Treat them with dignity. Do not carry people who can walk. Individuals may feel social stigma due to short stature. Counseling or support groups may be appropriate based on the unique needs of each child.

# Physical Activity, Trips, Events

## What you need to know

Exercise and physical education should be encouraged for strength building and obesity prevention. Choose activities to include children as appropriate if activity can be modified. Keep them safe and involved.

- 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
    - Swimming and biking are encouraged
- Care must be taken to limit risks for injury to the spinal cord due to decreased size of spinal column in the neck area. It is important to be aware of head and neck trauma. This includes avoiding:
    - Contact sports like Football, Hockey, or Rugby

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- Using trampolines
- 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. Assistive devices are available to improve accessibility and independence in all environments.

Field trip planning - 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

## School Absences and Fatigue

Students shouldn't have excess school absences. Coordinate with parents if any medical conditions cause the child to miss school.

Apnea may lead to daytime sleepiness

## Emergency Planning

### What you need to know

There should be a plan for an 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

# Resources

## Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide good medical overview of the condition without being too technical.
https://medlineplus.gov/genetics/condition/achondroplasia/

## Little People of America

Little People of America (LPA) is a nonprofit organization that provides support and information to people of short stature and their families. Click on the Resources tab and scroll down to "For Parents and Teachers" to find helpful tools to support school success.
https://www.lpaonline.org/

## Understanding Dwarfism

This program is dedicated to finding creative ways to change the world's perception of people with dwarfism. Through educating the general public, they want to create a brighter future for all those with dwarfism. http://understandingdwarfism.com/

## Dwarf Parents Blog

This is a blog for parents of children with dwarfism, however, there are some specific examples of accommodation requests for the school setting that may be helpful for other parents and school personnel. https://dwarfparents.com/category/school

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# Info for School Nurse and Primary Care Staff

Achondroplasia can present with multiple issues and will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

*Gene Reviews* is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry on achondroplasia provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. https://www.ncbi.nlm.nih.gov/books/NBK1152/

## HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

## Example letter for physician to provide to school

"My patient_____ has a diagnosis of achondroplasia. This condition necessitates an Individualized Education Program (IEP) or 504 plan.

In addition to a short stature, children with achondroplasia can have issues with hearing loss. Because of a narrow spinal canal, these children are more vulnerable to spinal cord injuries. Evaluation by OT can help identify what accommodations are most appropriate to minimize injuries and increase the student's comfort and ability to participate in daily classroom activities."

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# 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 always having a water bottle with her (she can overheat easily) or having some adaptive PE to prep her for regular gym classes. Although she must 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.

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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 she 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!

*Thank you to Laci-Mae and her family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.*