Achondroplasia
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.negenetics.org).

Physical characteristics and/or symptoms

Note: not all people with Achondroplasia syndrome will have all of these features.

- Disproportionate short stature, with disproportionately short limbs (growth charts: **Error! Hyperlink reference not valid.**)
    - Average height: men: 131cm
    - Average height women: 124cm
- Increased head circumference (see growth charts above)
- Intelligence is usually normal.
    - Unless hydrocephalus or other central nervous system complication is present
    - Gross motor development is different due to short limbs and large head Delays as an infant are common (Modified Denver: https://publications.aap.org/view-large/figure/8820700/PEDS_20201010_f2.jpeg)
    - Log rolling, seat scooting, snowplowing and reverse snowplowing are more common than typical crawling
- Obesity
    - In adults this can:
        - Contribute to morbidity associated with lumbar stenosis
        - Contribute to joint problems
        - Lead to possible cardiovascular complications
- Spine issues
    - Cervical spine impingement can sometimes occur in infancy
        - May present with arching or abnormal positioning
    - Kyphosis – presents in infancy
    - Lumbar lordosis – develops with upright posture
    - Spinal stenosis – develops as an adolescent or adult
        - Involves L1-L4

        - May present with numbness, weakness, and altered deep tendon reflexes.
        - May develop intermittent, reversible, exercise-induced limping **or may progress** to severe irreversible abnormalities of leg function
- Joint issues
    - Limitation of elbow extension and rotation
    - Excessive mobility of the knees, hips, and most other joint
- Sleep apnea
    - Can present early in life
- Middle ear dysfunction
    - If inadequately treated this can lead to hearing loss that may interfere with language development.
- Bowing of legs
    - Can be functional due to lax ligaments
    - Bowed tibia can develop due to overgrowth of fibulae
- Characteristic facial features
    - Frontal bossing or prominent forehead
    - Low nasal bridge - can cause increased incidence of nasal congestion
    - Midfacial hypoplasia - can contribute to sleep apnea

Recommended Routine Surveillance

- 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
- Monitor for sleep apnea
- Health Supervision Guidelines are available from the Academy of Pediatrics https://publications.aap.org/pediatrics/article/145/6/e20201010/76908/Health-Supervision-for-People-With-Achondroplasia

Emergency Protocols

- There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.
- Emergencies should be handled as with any child.
- Anesthesia poses a particular risk for individuals with achondroplasia and should only be undertaken by an anesthesiologist with particular experience

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8297512/pdf/cureus-0013-00000015832.pdf

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- ENT
  - Middle ear infections and monitor speech delays and hearing loss
  - Treatment of obstructive sleep apnea
- Geneticist / Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Orthopedics
  - Monitor growth and for presence of kyphosis
  - Monitor lower extremities for bowing
  - Treat tibia bowing as needed
- Neurology
  - Hydrocephalus and/or craniocervical junction compression may be present
  - May require involvement of a neurosurgeon for a decompression surgery
- Developmental Evaluation with OT/PT/ST
  - Adaptive equipment and strategies
  - Hypotonia
  - Speech issues may be present

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school

My patient_____ has been diagnosed with Achondroplasia. Achondroplasia is the most common form of short-limbed dwarfism. Although the inheritance is autosomal dominant, most children with achondroplasia will be born to average stature parents. The typical features include short stature, an average sized trunk, short limbs with more pronounced shortening of the upper arms and thighs, limited range of motion at the elbows and large heads. Medical complications with

achondroplasia include apnea, obesity, recurrent ear infections, and curvature of the spine. Narrowing of the lower spinal canal causing pain and numbness can occur. Instability at the top of the spine can also occur, requiring care in activities that put strain on the neck. Because of these, _____ needs the following accommodations.

- Sample letters for parents to send to schools to explain the condition: https://lpa.memberclicks.net/assets/school%20letters%202016.pdf

Seven Important Aspects of School Life

"Achondroplasia at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

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

GeneReviews: National Center for Biotechnology Information (NCBI) Bookshelf - Achondroplasia
http://www.ncbi.nlm.nih.gov/books/NBK1152/

Little People of America
http://www.lpaonline.org/