Potential Complications Associated With ACHONDROPLASIA

intheknow ABOUT ACHONDROPLASIA

The American Academy of Pediatrics (AAP) published a report in June 2020 to provide guidance to healthcare providers on how to monitor for complications associated with achondroplasia. This is a summary of the report and is not comprehensive. It can introduce you to some of the complications or symptoms your healthcare team may be watching for and can help in discussions when deciding the right care.

Keep in mind that everyone's experience with achondroplasia is different. This summary is not a substitute for medical advice. Raise any questions you have with your healthcare team to manage any healthcare needs.

The information is organized by age ranges to help guide you based on where you are in your journey. *(Select an age group on the right to learn more.)*

INFANT *(BIRTH TO 1 YEAR OF AGE)*

CHILD *(1 YEAR TO 13 YEARS OF AGE)*

TEEN & YOUNG ADULT *(13 YEARS TO 21 YEARS OF AGE)*

ADULT *(21 YEARS OF AGE AND OLDER)*

INFANT
(BIRTH TO 1 YEAR OF AGE)

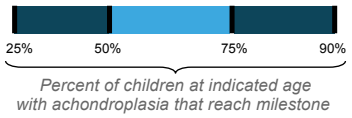
In the first year of life, infants need extra attention and supervision. The healthcare team may monitor for nervous system and breathing complications.

POTENTIAL COMPLICATION/ FUNCTIONAL CONSEQUENCE	WHAT IT LOOKS LIKE	WHAT CAN BE DONE
Excessive outward curve of the spine, also known as thoracolumbar kyphosis *(caused by weak torso muscles)*	• Forward bending of the spine • Motor delays	• Back support should be provided while sitting **Should self-resolve as child supports their weight**
Ear infection, also known as otitis media	• Language delays • Hearing loss • Ear pain	• Hearing tests should be done by 9 to 12 months, and be part of each visit
Breathing problems *(caused by how the bones in the face grow, small chest size, and smaller lung volume)*	• Choking or gagging while feeding • Pauses in breathing during sleep • Lips turning blue while eating or sleeping	• Routine monitoring is recommended (until age 8) • Surgery may be required; for example, the removal of tonsils and/or adenoids
Buildup of fluid on the brain, also known as hydrocephalus	• Bulging of the head • Forehead is hard to the touch • Sleepiness • Irritable • Poor weight gain • Marked developmental delay	• Head growth with achondroplasia-specific growth charts measured at every visit • Neuroimaging (eg, MRI), if it can be done safely
Squeezing of the spinal cord where the skull meets the spine (also known as craniocervical junction constriction), includes: • Narrowing of the foramen magnum (hole at base of the skull) • Disrupted brain signals to control breathing (central sleep apnea) • Change in shape of the upper spinal cord • Lack of fluid around the spinal cord	• Choking or gagging while feeding • Pauses in breathing during sleep • Lips turning blue while eating or sleeping • Weak muscle tone or "floppiness" • Asymmetric reflexes • Muscle spasms in legs • Poor weight gain, even though feeding is sufficient	• Overnight sleep study • Neuroimaging (eg, MRI), if it can be done safely

DEVELOPMENTAL MILESTONES
(BIRTH TO AROUND 4 YEARS OF AGE)

While it may take a bit longer, motor and language milestones will likely happen for children with achondroplasia **in their own time**. It is important that timing of milestones should only be compared amongst children with achondroplasia. In the same way, **height and weight should be plotted on achondroplasia specific growth charts**.

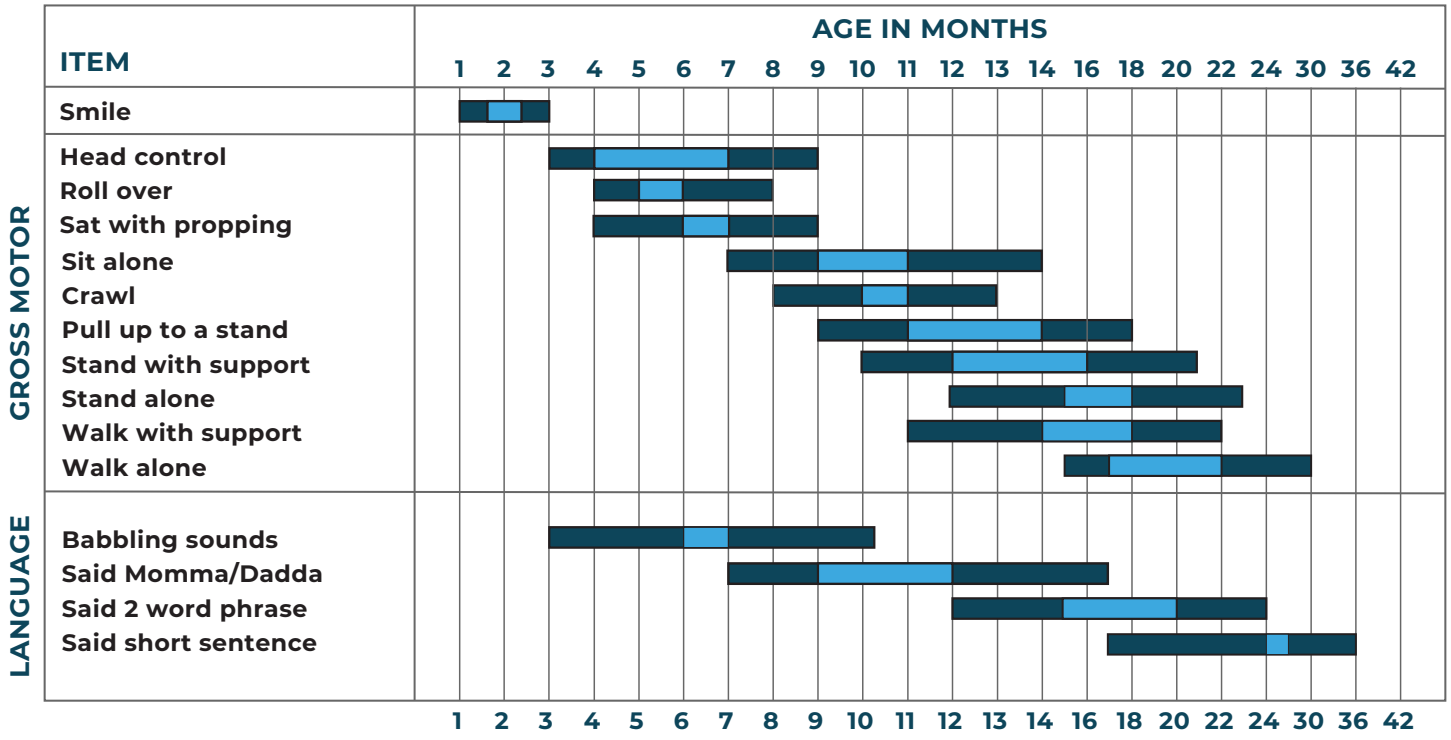
How to read the graph below:



Percent of children at indicated age with achondroplasia that reach milestone

For example, when looking at ***Smile***, 25% of children with achondroplasia are able to smile when they are 1 month old, and 90% are able to smile by the time they are 3 months old.

Ages when children with achondroplasia reach developmental milestones



RECOMMENDATIONS
AS THESE MILESTONES ARE ACHIEVED

You can help protect your infant/child by following some basic recommendations, like those below. Ask your doctor for recommendations for the best types of car seats and other supportive devices for your child.

DO's

- **Support** head, neck, and full spine
- Use a **solid-back (no excess padding)** stroller
- Keep in a **rear-facing car seat** as long as possible

DON'Ts

- Do not let them sit **unsupported**, which could allow **excessive curving** of the back (kyphosis). Avoid **"umbrella-style" strollers**
- Do not use **mechanical swings** before the baby has head control and can keep airway open. Avoid **carrying slings** at all times

CHILD
(1 YEAR TO 13 YEARS OF AGE)

Be sure to ask your doctor about any concerns you have about your child's development. Remember: Achondroplasia does not affect your child's cognitive development (intelligence).

POTENTIAL COMPLICATION/ FUNCTIONAL CONSEQUENCE	WHAT IT LOOKS LIKE	WHAT CAN BE DONE
"Sway back," also known as lumbar lordosis	• Inward curving of the lower back • Walking and supporting of their own weight may be delayed (but these milestones are expected to be reached by 2 to 2.5 years old)	• Monitor for decreased joint motion due to shortening of muscle, tendon, or other tissue • Gentle stretching with the help of a physical therapist or pediatric orthopedist
Leg bowing, also known as genu varum • Changes to the cartilage and bone surrounding the knee • Shinbone (tibia) and bones between knee and ankle (fibular) are bowed, or curved	• Outward bowing of legs • Pain • Difficulty walking	• Monitor for knee instability while walking, pain, and effects on daily activities • A pediatric orthopedist is seen if problems occur
Obstructive sleep apnea *(caused by partial or complete blocking of the airway while sleeping)*	• Increased snoring • Pauses in breathing during sleep • Choking and/or deep sighs • Bed wetting • Vomiting • Excessive sweating	• Overnight sleep study
Hearing impairment	• Language delays	• Hearing tests should be done every year • Speech evaluation should be done no later than 2 years of age
Acid reflux, also known as gastroesophageal reflux	• Burning sensation in chest, especially after eating • Chest pain or sensation of lump in your throat • Difficulty swallowing	• Potentially change types of food being eaten and eating habits • Consult with a pediatric gastroenterologist or pulmonologist
Obesity *(mid to late childhood)*	• Weight gain	• Monitor weight and eating habits • Recommend the child stays active
Dental issues	• Overcrowding of teeth	• Braces or palate expansion might be needed • Dental extractions are common
Narrowing of spaces in the vertebrae, which surrounds and protects the spine, also known as spinal stenosis	• Lower back and/or leg pain • Weakness or numbness in the legs after walking • Difficulty controlling bladder function	• Deep tendon reflexes need to be checked yearly for asymmetry or increased reflexes • May require surgery

TEEN & YOUNG ADULT
(13 YEARS TO 21 YEARS OF AGE)

Emotions. Peer pressure. The need to fit in. The teenage years are difficult for everyone. But for a teenager with achondroplasia, this time of life can be even harder.

In addition to the potential complications listed to the right *(click to learn more)*, pain as well as feelings of depression, anxiety, and isolation are common. Emotional support is critical during these years.

- Spinal stenosis
- Dental issues
- Obesity
- Obstructive sleep apnea
- Hearing impairment

POTENTIAL COMPLICATION/ FUNCTIONAL CONSEQUENCE	WHAT IT LOOKS LIKE	WHAT CAN BE DONE
Pain	• Back and leg pain related to spinal stenosis • Inability to perform activities of daily living or desired physical activity	• Assess for pain and its effects on activities of daily living and desired weight control
Psychosocial impact	• Barriers to equal opportunity in education and employment • Social isolation • Comfort in one's identity	• Review social adaptation and discuss with your child, his or her friends, and teachers about short stature • Encourage participation in social activities and support groups • Set career and life goals high and appropriate

ADULT
(21 YEARS OF AGE AND OLDER)

Adults may outgrow some complications, while other complications may continue or new complications may arise. Mobility and pain can continue to be challenging, affecting work, physical activity, and even daily household responsibilities.

Health recommendations for adults with achondroplasia are specific to their symptoms. **The most common complication** occurring in adulthood is related to **spinal stenosis.**

FAMILY PLANNING CONSIDERATIONS

The opportunity to conceive and have children is no different for women with achondroplasia than for those of average height. Females, who **would like to conceive**, could:

- Seek genetic counseling before pregnancy since achondroplasia is a genetic condition that can be passed down to children. The risk of passing on conditions, and the outcomes of doing so, are different if both parents are affected
- Establish where the infant will be delivered if a genetic condition is suspected. This center should have the appropriate services available in the event complications arise
- Consult with a high-risk maternal or fetal medicine specialist to discuss delivery options for a cesarean section, which a female with achondroplasia will undergo because of their smaller-sized pelvis

KNOWING WHAT TO EXPECT CAN HELP CAREGIVERS AND PEOPLE WITH ACHONDROPLASIA PLAN AND PREPARE FOR THE FUTURE

Use your phone's camera to scan and download the **Care Team Guide** and learn about the specialists who can help manage achondroplasia.

Hear stories from families who have been right where you are.

https://www.achondroplasia.com/youre-not-alone-in-achondroplasia/

Contents from this resource have been adapted from Hoover-Fong J et al. Health supervision for people with achondroplasia. Published in *American Academy of Pediatrics,* June 2020.