together

The achondroplasia journey from diagnosis to 18 years of age

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Introduction

Introduction

Acknowledgements

BIOMARIN

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STOWARZYSZENIE NA RZECZ DZIECI ODBLOKUJ ŻYCIE Z ACHONDROPLAZJĄ

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The Achondroplasia Roadmap

This Roadmap is for you if you have – or will have – a child with achondroplasia. It will help you to prepare for the road ahead, wherever you are on your journey.

For each stage of your child's development, you will find information about:

- **YOU:** What you and your child can expect at that stage
- **HEALTHCARE:** How to support your child's healthcare at that stage
- **SOCIAL:** How you or your child may want to discuss achondroplasia – and the ways it impacts you – with the people around you

This Roadmap will also help you understand the possible ways to care for achondroplasia and which health professionals may be involved. There are also links for further information and support.

You can work your way through the roadmap at your own pace. If you want to go back to the start, click the **"HUB"** icon at the bottom of any page.

If your questions are not answered, it is a good idea to speak with your child's healthcare team. Or, you could reach out to a group dedicated to achondroplasia, known as an achondroplasia patient association.

The content of this roadmap has been written with the support of people with achondroplasia and their parents and caregivers.

What is achondroplasia?

Achondroplasia is a rare condition caused by changes in a gene that controls physical growth. This results in short arms and legs (referred to as 'disproportionate short stature') and a relatively large head, with an average-sized torso. The child's short stature will remain as they reach their teenage years and adulthood.[1]

Achondroplasia affects 1 in 20,000 people.[1] Most people with the condition have parents of average height. If one or both parents have it, the chances of having a child with achondroplasia are higher. Many children with achondroplasia live full and healthy lives, with their intelligence and life span not typically impacted by their condition. They have some delayed physical development as a child.[1]

Hub

Hub

You

It's our journey: let's see what our family can expect and what action we can take

Healthcare

Let's discuss health concerns associated with achondroplasia and the decisions you may need to make

Social

Let's see how social aspects of achondroplasia will affect your family over time

Before birth (pre-natal)

1. Diagnosis of achondroplasia before birth
2. Genetic counseling
3. Pregnancy, birth and postpartum (after birth)

Birth to age 2

4. Diagnosis of achondroplasia when the child is born
5. Learning how to care for a child with achondroplasia
6. Child starts to move, walk and talk

Ages 3 to 6

7. Child becomes independent of parents
8. Child starts to develop self-awareness
9. Child starts nursery or preschool
10. Child starts to make friends
11. Child starts primary school

Ages 7 to 12

12. Child gains more awareness of achondroplasia
13. Child starts secondary school

Ages 13 to 18

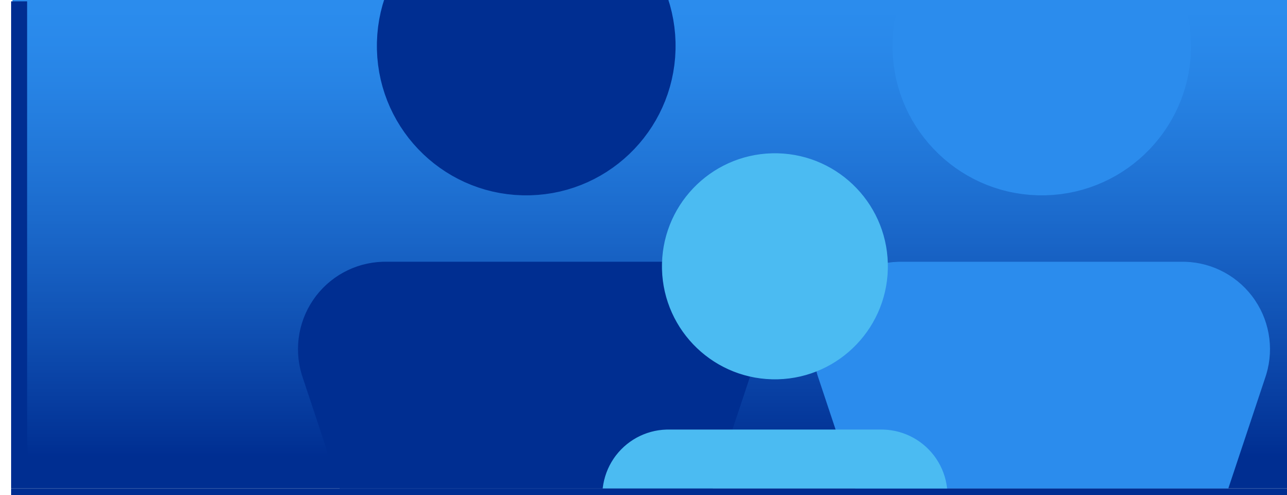
14. Young person's sense of self-awareness develops further
15. Young person begins to seek teenage independence
16. Sex and relationships
17. Transition to adult healthcare

Healthcare Considerations

18. Between birth and age 2
19. Between ages 3 and 6
20. Between ages 7 and 12
21. Between ages 13 and 18

Additional Resources

References



1

Diagnosis of achondroplasia before birth

Before birth (pre-natal)

You

Healthcare

Social

What can parents expect at this stage?

Achondroplasia may be picked up during routine scans in the third trimester of pregnancy (from 28 weeks).[2] These scans may show potential signs of the condition, e.g., shorter arms or fingers.[3] Achondroplasia happens in around 1 in 20,000 babies. Most children with achondroplasia have average-size parents.[1]

You may feel overwhelmed at hearing this diagnosis. It is reassuring to know that children with achondroplasia can live a full and healthy life, with their intelligence and life span not typically impacted by their condition.[1] Understand that there is nothing you did as parents to cause your child's achondroplasia.

Each person's experience with achondroplasia is different. With your love and support, life with a child who has achondroplasia can be one of joy and happiness.

Advice for parents

It is useful to know where to find trustworthy information about achondroplasia. You could:

- Use information in this Roadmap.
- Look at achondroplasia patient association websites.
- Speak to your doctor about what to expect when your child is born.

Other children in your family may need emotional support and extra attention during this time. You may wish to:

- Let siblings know that their new brother or sister will have achondroplasia.
- Bring them with you to appointments so they can ask questions.
- Look at achondroplasia patient association websites together.

Achondroplasia patient associations are a great way to meet people who have experience with the condition or who live with it themselves.

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1

Diagnosis of achondroplasia before birth

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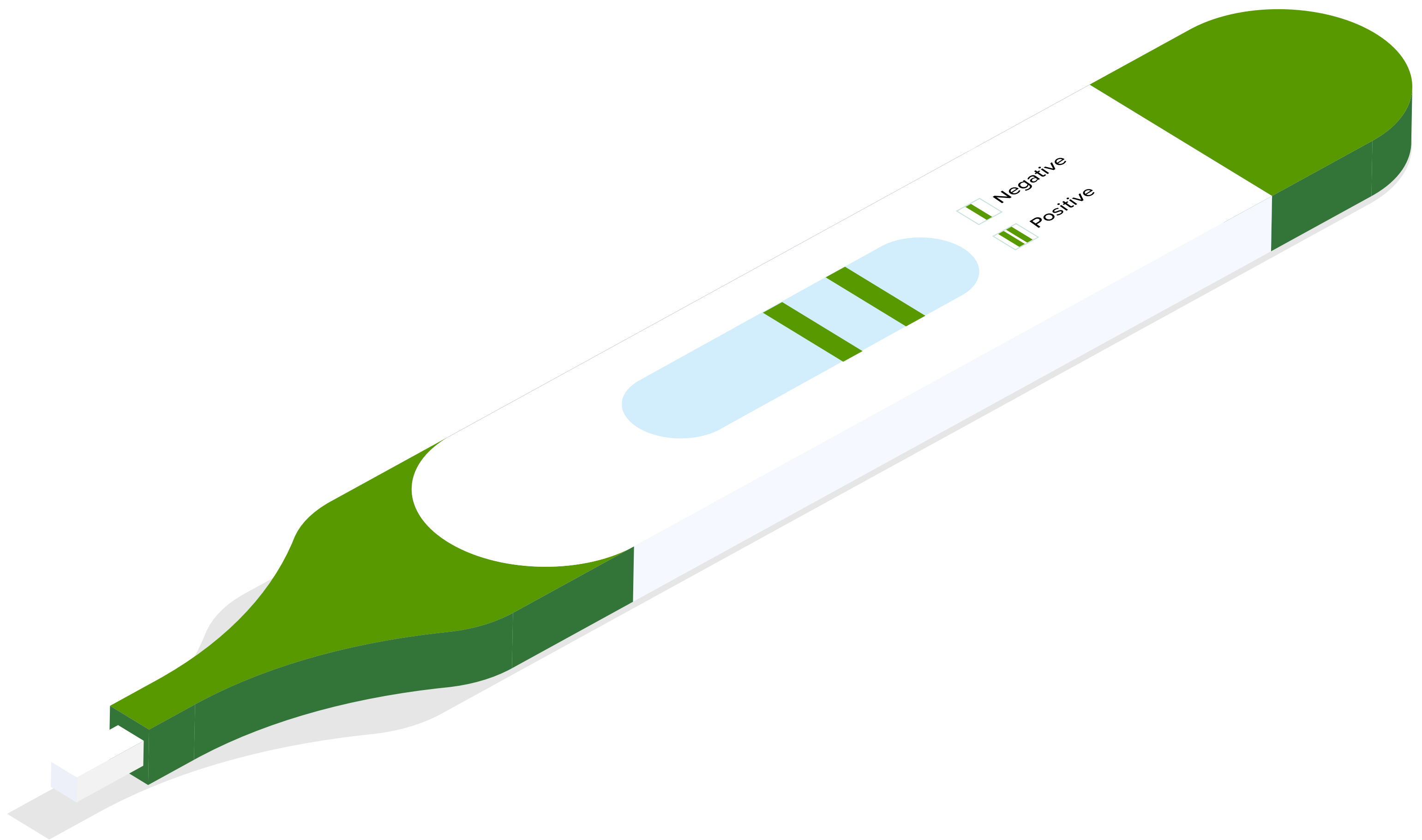
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

You should be able to meet with specialist doctors to discuss how to care for a child with achondroplasia and ask any questions.

Questions to ask your healthcare team

- Why does my child have achondroplasia?
- What will achondroplasia mean for my child when they are born?
- If I have other children in the future, will they have achondroplasia?
- What are the differences between a child with and without achondroplasia?
- Will my child still be able to live a full, healthy life?
- What are the risks of giving birth to a child with achondroplasia?
- Who will look after me during pregnancy and birth?



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Diagnosis of achondroplasia before birth

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Birth to age 2
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Ages 13 to 18
Healthcare considerations

How to describe achondroplasia to people around you

Your friends and family may have questions about achondroplasia. Only talk about it when you feel ready.

You could say:

- Achondroplasia is a rare condition.
- Your child will have a noticeably short stature.
- They will be able to live a full and healthy life.
- They may need some procedures and tests during their life.

You can decide what words to describe achondroplasia are most comfortable for you, then ask people to use the wording you prefer. This will help prepare a healthy and happy social environment for your child.

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Genetic counseling

Before birth (pre-natal)

You

Healthcare

Social

1. Diagnosis of achondroplasia before birth
2. Genetic counseling
3. Pregnancy, birth and postpartum (after birth)

What can parents expect at this stage?

Achondroplasia is a genetic condition, so genetic counseling may be offered to you.[1,4] Genetic counseling will give you information about how achondroplasia could affect you or your family.

You can have genetic counseling when you're planning a family, during pregnancy or after your child is born.[4,5] Read more about genetic counselling in the **'Healthcare'** section.

How does this differ for parents with achondroplasia?

It is recommended that people with achondroplasia speak with a genetic counsellor if they are planning a family. This is because the chance of having a child with achondroplasia is increased. This also gives your healthcare team time to plan for pregnancy and birth.[6]

Advice for parents

You may want to write a list of questions to ask your genetic counselor (find suggested questions in the **'Healthcare'** section). This will help them focus on what's important to or worrying you.

Genetic counseling involves looking at your family history of medical conditions, so you may want to think about this beforehand.[4,5]

One important role of the counselor is to give you emotional support, so share how you are feeling with them. You can follow-up with questions, doubts, and anxieties you may have during your pregnancy.

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2

Genetic counseling

Before birth (pre-natal)

You

Healthcare

Social

1. Diagnosis of achondroplasia before birth
2. Genetic counseling
3. Pregnancy, birth and postpartum (after birth)

Your genetic counselor may ask questions about:

- Your own health.
- Your partner’s and wider family’s health.
- Your ethnic background.

These questions help the counselor understand your family medical history and any conditions your child might be more likely to have.

Together, you may explore options for:

- Possible tests you could have.
- Managing your pregnancy and labor.
- Planning future pregnancies, if this is a concern.

Questions to ask your genetic counsellor

- Can you explain why our child has achondroplasia?
- Does achondroplasia run in families?
- If either my partner or I have achondroplasia, are our children at risk of having it?
- What kind of genetic tests are available and how are they done?
- What kind of information can testing give me?

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Genetic counseling

How to talk to about genetic counseling

It is up to you who you talk to about genetic counseling and how much you tell them.

Family

You may want to speak to your wider family about their own medical history. For example, you may need to ask them about conditions that run in the family.

Work

You may want to arrange a confidential chat with your company's HR team or your manager, so that a plan can be made to support you. For example, to arrange time off for genetic counseling appointments or additional tests.

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Pregnancy, birth and postpartum

Before birth (pre-natal)

You

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Social

1. Diagnosis of achondroplasia before birth
2. Genetic counseling
3. Pregnancy, birth and postpartum (after birth)

What can parents expect at this stage?

As with any pregnancy and birth, understanding what might happen, and about your choices, will help you to feel more relaxed and confident. Read more in the **‘Healthcare’** section.

Advice for parents

Having a new baby can be hard. You may want to seek support from your healthcare team to discuss any worries or negative feelings.

You may need to ask for extra support from family and friends if mother and baby stay in hospital (e.g., following a cesarean), especially if you have other children. It is a good idea to arrange this support before the birth.

How does this differ for parents with achondroplasia?

If you are a person with achondroplasia, there are differences in how you are cared for during pregnancy, birth and after your child is born. Read more in the **‘Healthcare’** section.

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3 Pregnancy, birth and postpartum

Before birth (pre-natal)

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Birth to age 2
Ages 3 to 6
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Ages 13 to 18
Healthcare considerations

If your child has been diagnosed with achondroplasia

Pregnancy

In some pregnancies, there may be too much fluid around the baby in the womb. This is called polyhydramnios.[7,8] This is usually seen during scans towards the end of the pregnancy. If this happens, you can discuss how to treat it with your healthcare team.

Giving birth

Cesarean delivery (where the baby is born during an operation) is recommended because of the baby's head size and shape. Use of forceps will be avoided, because they place too much stress on the baby's spine.[7]

After birth

You may be referred to a hospital with expertise in looking after babies with achondroplasia. Access to these hospitals varies depending on where you live. Achondroplasia patient associations might be able to help you find a specialist team closer to you.

Some babies with achondroplasia have trouble breathing at birth.[7] They may need to go straight to the intensive care unit. While this can be worrying, this means your baby's breathing can be checked and they can be helped if necessary.

If you are a mother with achondroplasia

Pregnancy

Your care should be with an obstetrician (a doctor who specialises in care during pregnancy, labour and after birth) and not with the midwife team alone. This is so the pregnancy can be closely monitored. This also allows you to prepare for a cesarean delivery.

During pregnancy:[6]

- You should be offered scans every 8 weeks or so. This varies according to where you live.
- Weight gain is common, which can lead to back pain. You can discuss ways to manage this with your healthcare team.
- There is limited space inside your body for your baby to grow. In the last 12 weeks of pregnancy, your baby may put more pressure on your lungs and you may find it harder to breathe.
- You should discuss what pain relief options are available for birth and decide what is best for you.

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3

Pregnancy, birth and postpartum (cont.)

Before birth (pre-natal)

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Giving birth

A cesarean delivery (where the baby is born during an operation) is most common for people with achondroplasia. A vaginal delivery is usually difficult if you have a smaller body frame, especially after 32 weeks of pregnancy.[6,7]

It's more common to have your baby before full-term (40 weeks). Often, this will be recommended by your doctor when you talk about your options at the start of pregnancy, giving you time to prepare.[6,7]

If you have spinal issues, it can be difficult to have a type of pain relief called an epidural. This is because it is injected in the spine area. In this case, a general anesthetic will be recommended ('being put to sleep').[7]

After birth

Your care should be in an environment that allows you to easily look after your baby and recover from birth. This means:

- An appropriate height bed for you.
- A bedside cot for your baby at the right height.
- Easy access to toilets and washrooms.

You can discuss this with your healthcare team before the birth. If you need extra help after your baby has been born, let one of your healthcare team know.

Questions to ask your healthcare team

- How regularly will I have ultrasound scans during my pregnancy?
- What are my pain relief options for birth?
- What type of birth is recommended for me and why?
- What sort of specialist care might my baby need after birth? Are these facilities available in this hospital?
- What experience do you have delivering a baby with achondroplasia / delivering a baby for a mother with achondroplasia?

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Pregnancy, birth and postpartum

Speaking to people after the birth

Only let visits happen when you feel ready. It may be the birth partner's job to be 'gatekeeper'.

When you do introduce your child to family and friends, you may want to start having conversations about achondroplasia straight away. Or, you may wish to wait until after the first visits to discuss achondroplasia.[1]

4

Diagnosis of achondroplasia when the child is born

You
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Before birth (pre-natal)
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What can parents expect at this stage?

Achondroplasia is a genetic condition that results in short stature. Achondroplasia is usually diagnosed during pregnancy, but sometimes the signs aren't seen until the child is born. It happens in around 1 in 20,000 babies born. Most children with achondroplasia have average-size parents.[1]

You may feel overwhelmed by your child's diagnosis. It is probably not what you expected. It is reassuring to know that children with achondroplasia can live full and healthy lives.

Each person's experience with achondroplasia is different. With your love and support, life with a child who has achondroplasia can be one of joy and happiness. Understand that there is nothing you did as parents to cause your child's achondroplasia.

Advice for parents

It is useful to know where to find trustworthy information about achondroplasia. You could use information in this roadmap or look at achondroplasia patient association websites.

Other children in your family may need emotional support and extra attention during this time. You may wish to:

- Let siblings know that their new brother or sister will have achondroplasia.
- Bring them with you to appointments so they can ask questions.
- Look at achondroplasia patient websites together.

If you need support, it may be helpful to speak with:

- Your doctor.
- Psychological support.
- An achondroplasia patient association..

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4

Diagnosis of achondroplasia when the child is born

Birth to age 2

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Before birth (pre-natal)
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Ages 7 to 12
Ages 13 to 18
Healthcare considerations

You will meet with several different healthcare professionals who help you prepare for life with your new baby.

You may be referred to a hospital that specialises in managing children with achondroplasia. Access to these hospitals will vary depending on where you live. Achondroplasia patient associations might be able to help you find a specialist team.

Professionals who may be in your healthcare team include:

- Anesthetist
- Genetic counselor
- Geneticist
- Neonatologist
- Nursing team
- Obstetrician
- Gynecologist
- Occupational therapist
- Orthodontist
- Otorhinolaryngologist (Ear, Nose and Throat specialist)
- Pediatric orthopedic specialist
- Physiotherapist
- Primary care physician
- Psychologist
- Neurosurgeon
- Speech-language pathologist
- Dietitian

Questions you may want to ask your healthcare team

- Why does my child have achondroplasia?
- What does achondroplasia mean for my child?
- If I have other children, will they have achondroplasia?
- What are the differences between a child with and without achondroplasia?
- Will my child still be able to live a full, healthy life?
- What are the risks of giving birth to a child with achondroplasia for the first time?
- Who is in charge of what aspects of the care of my child?
- What are my/my family’s options for emotional support?

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Diagnosis of achondroplasia when the child is born

Talking about your child's achondroplasia diagnosis

You may find it helpful to prepare a simple explanation of achondroplasia. In particular, you may want to think about how you will talk to other siblings about achondroplasia.

You could say:[1]

- Achondroplasia is a rare condition.
- Your child will have a short stature.
- They will be able to live a healthy and typical life.
- They may need some procedures and tests during their life.

You can decide what words to describe achondroplasia are most comfortable for you, then ask people to use the wording you prefer. This will help prepare a healthy and happy social environment for your child.

5

Learning how to care for a child with achondroplasia

What can parents expect at this stage?

During the first two years, you will adapt to life with a child with achondroplasia. This includes learning about their physical development and how to manage any additional health needs.[6]

Advice for parents

Take time to reflect on how you and other members of your family are feeling. If you are struggling with caring for your child, you may want to:

- Talk to a healthcare professional or achondroplasia patient association.
- Join in with achondroplasia patient association community events, activities or awareness campaigns.

Sometimes, people may react negatively to your child's short stature.[9] You may want to discuss with your partner and family how you will deal with such situations. Read more in the **'Social'** section of this chapter.

Practical matters

There may be some practical matters to manage during this time. You may want to:

- Talk to your employer if you need to take time off work to take your child to appointments.[9] Ask your employer if any support is available through the workplace.
- Explore financial support, including social security benefits. These vary by country and for each family's circumstances. Speak with a local achondroplasia patient association for advice.
- Seek a disability certification for your child. This helps to secure benefits such as parental leave, social security and tax benefits, educational, housing, economic, and health benefits. The process will vary by country, so reach out to your local achondroplasia patient association for advice.

Breastfeeding

Some babies with achondroplasia find it hard to breastfeed.[10] If this happens, you may want to speak with a breastfeeding counsellor who can help with technique or other feeding options.

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Learning how to care for a child with achondroplasia (cont.)

Birth to age 2

You

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Social

Before birth (pre-natal)
Birth to age 2
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Ages 13 to 18
Healthcare considerations

Promoting safe sleep

Some children with achondroplasia have a prominent back of the head and, if placed on their back for sleep, their head will be bent forwards. This can cause breathing problems.[11]

Speak with your healthcare professional regarding a safe position for your infant to sleep. There is guidance suggesting you could use a special cushion, tightly rolled towel or washcloth behind their neck.[12,13]

Older infants with achondroplasia (6 months and older) may choose to sleep on their back with their neck extended and face to the side. They usually do this to help them breathe more easily.[13]

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5

Learning how to care for a child with achondroplasia

During their early years, your child will likely spend a lot of time in healthcare environments. You may want to:

- Find out more about achondroplasia so you can inform doctors about your child's needs. Resources like this Roadmap may help, as well as resources from achondroplasia patient associations.
- Raise any concerns you have about your child's health or development, which may not be obvious to their healthcare team.
- Explore some of the treatments that may be available to help your child's physical development.
- Contact an achondroplasia patient association for advice on tracking your child's development. Development charts are available for children with achondroplasia.[6]

Testing for common physical issues

Your child's healthcare team will begin to do regular tests and monitoring for signs of physical problems that are more common in children with achondroplasia. For example:[6,10,14]

- Spinal issues
- Hearing issues
- Sleep apnea

Preventing spinal issues

Children with achondroplasia often have a curve in their back, which can cause spinal issues. This usually goes away as they get stronger and begin to walk.[6] There are things you can do to help prevent this:

- When lifting or carrying your newborn baby, support their head and neck (like any baby). For babies with achondroplasia, it is also important to support their lower back whenever lifting or carrying.[10,15]
- When winding your child, avoid placing them in a seated position. Try keeping your baby upright against your chest.[15]
- Avoid 'unsupported sitting' too early, as it puts their spine into a C-shaped curve. It is recommended you place your baby on their tummy, side, or back when they're awake. You can let your child sit in a C-shaped curve position once they're able to move themselves into a sitting position.[15]
- Avoid soft canvas versions of things like baby rockers and bouncers, baby walkers and strollers. They don't give your child enough back support and may put a strain on their neck. Avoid swings and trampolines for the same reason.[10,11,15]
- Avoid baby carriers and baby wraps as they open the child's legs and can damage the hips.[10]

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4. Diagnosis of achondroplasia when the child is born
5. Learning how to care for a child with achondroplasia
6. Child starts to move, walk and talk

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Learning how to care for a child with achondroplasia (cont.)

- Look for appropriate car seats that allow babies to lie as flat as possible.[10,15] Car seat laws vary by country or state, so you might find it helpful to ask an achondroplasia patient association, your healthcare team, or parents of other children with achondroplasia for advice.

Questions to ask your healthcare team

- Do you have experience treating a child with achondroplasia?
- How can we work together to make sure my child has the best possible care?
- What therapies (for example, physiotherapy, aquatic therapy) are available for my child?
- What treatments or procedures might my child need?
- Why does my child need so many tests? And what are they looking for?
- What signs and symptoms should I look out for at home?

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Learning how to care for a child with achondroplasia

Dealing with negative social situations

Sometimes, people may react negatively or say hurtful things about your child's short stature. This can be distressing for you and your family. You may:

- Ignore anybody reacting negatively or saying hurtful things.
- Remove yourself and your child from the situation.
- Talk or explain to the person.

People may be unaware that something they have said is inappropriate, particularly other children.

Speaking with other parents or families who have a child with achondroplasia can help you feel connected to a community of people who understand the challenges you face.

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Child starts to move, walk and talk

Birth to age 2

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5. Learn how to care for a child with achondroplasia
6. Child starts to move, walk and talk

Before birth (pre-natal)

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Ages 13 to 18

Healthcare considerations

What can parents expect at this stage?

Your child will continue to develop and become more mobile and independent.

Children with achondroplasia may do things like rolling over, sitting up, standing, and walking later than children of average stature.[6] They will reach these milestones when they are physically ready. Children with achondroplasia usually sit up alone from around 12 months.[11] Standing alone at about 16–29 months is common.[15]

For children with achondroplasia, movement and walking development may be affected by several things:

- Nearly half of children with achondroplasia have bowed legs, where their legs curve outwards between the thighs. This can be painful.[16]
- The muscles and ligaments around the knee joint, which hold the knee in place and allow it to move, can be unstable.[11]
- Children with achondroplasia may be overweight, although it is not fully understood why.[16]

At first, your child may prefer to move around in other ways, such as ‘snow ploughing’, where the head is used as a balancing support. These movements are important milestones, showing they are adapting to their bodies. They will not harm your child or impact their ability to walk in the long-term.[10]

Your child may have developed a small ‘hump’ near their shoulders when they were a small baby (known as kyphosis). This may go away once your child begins to walk, although it does continue in some children.[16]

Some children with achondroplasia have difficulty learning to talk, due to:

- Problems with hearing, caused by repeated ear infections.[16]
- Problems related to the size / proportions of the tongue and mouth.[10,17]

Advice for parents

Try not to compare your child’s physical development to other children. Celebrate their first words and steps whenever they happen.

Trips and falls are common when your child starts to walk, although you can still encourage them to keep trying. You can buy safety ‘corner bumpers’ for your furniture to keep your child safe from sharp corners. Avoid baby walkers as they do not give enough support to your child’s back and neck.[18]

Try to encourage regular exercise.

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Child starts to move, walk and talk

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Help with walking development

If your child is struggling to walk, or certain movements are painful, it may be useful to speak with a physiotherapist or occupational therapist.[18] They can provide advice on how to help your child overcome these difficulties.

Help with speech development

Your child will have regular hearing tests and will see an ear, nose and throat specialist.[19] If your child's hearing is impacted by ear infections, your doctor might suggest putting in ear tubes. These can lower the chances of infection.[20]

Speech therapy can help test and treat your child for issues with their speech and general communication. Seeing a speech and language therapist as early as possible can help prevent and minimize any delays in learning to speak.[10,17]

Questions to ask your healthcare team

- I would like to track my child's physical development – what tools are available to help me?
- What can I do to help prevent spinal issues?
- What signs should I look out for in case my child has difficulty hearing?
- What sort of anesthetic will my child have if they need a procedure to put in ear tubes?
- Can you help me meet with an ear, nose and throat specialist?
- Does my child need to be tested by a speech-language therapist?

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Child starts to move, walk and talk

Talking about your child’s physical development

You may want to speak to family and friends about achondroplasia and what it means for your child’s physical development.

Remind people that all children reach milestones at different times. Some will need more support than others. Encourage people to not put pressure on your child to do things like walking and talking before they’re ready.

Child becomes independent of parents

What can parents expect at this stage?

Your child will begin to become more aware of the world around them and their place in it. They will start to develop new skills and take on tasks independently. For example:[12]

- Toileting
- Dressing
- Eating and drinking
- Playing, alone or with other children.

Children with achondroplasia may find it hard to do certain things on their own. Each child will reach these milestones at different times.[10]

Advice for parents

You can explore adaptations that could support your child to be more independent. For example, in helping around the house (see Healthcare section).

Try to balance providing help with encouraging independence. Stick with the process even if your child faces difficulties.

Ask your child to make some decisions for themselves, as appropriate. Do this in both healthcare settings and in your home, so that your child feels they have a say in different aspects of their lives.

Child becomes independent of parents

Your child may begin to resist tests or treatments that make them feel scared. Try to reassure your child and explain procedures as simply as possible. How you do this will depend on their age and maturity. You could:

- Let your healthcare team know, so they can support you in creating a calm atmosphere.
- Ask for advice from a specialist in child psychology on how to have these conversations. Check with your healthcare team to see if this is possible where you live.

Speak with an occupational therapist, physiotherapist or achondroplasia patient association about adaptations to help your child be as independent as possible. This may include things like:[21,22]

- Adaptations in underwear and clothing so that they can dress themselves, such as elastics, Velcro, and magnetic buttons.
- Lowered light switches or remote light switches.
- Home furniture adaptations (adjustable chair with back support, lowered bed, lowered closest rods, toys stored at a manageable height).
- Door openers and step stools.

Ages 3 to 6

You

Healthcare

Social

7. Child becomes independent of parents
8. Child starts to develop self-awareness
9. Child starts nursery or preschool
10. Child starts to make friends
11. Child starts primary school

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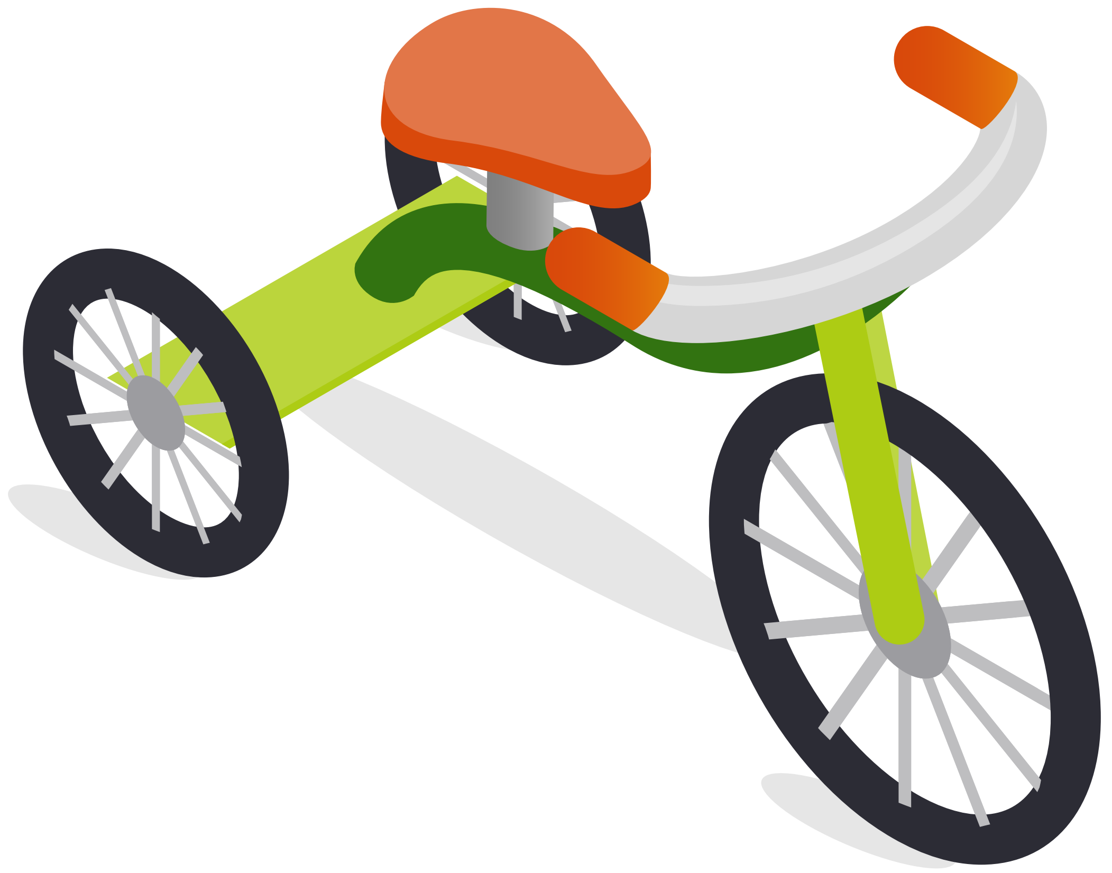
7

Child becomes independent of parents

Talking to your child about achondroplasia

As your child becomes more independent, you may want to talk to them about their achondroplasia and what this means for them. You could use online resources or contact an achondroplasia patient association for books written to explain achondroplasia to young children. Children’s achondroplasia booklets can be found in the additional resources section on page 68 of the Roadmap.



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You
Healthcare
Social

8

Child starts to develop self-awareness

What can parents expect at this stage?

At some point, your child will become aware that they are different from others. For most children with achondroplasia, this will be a gradual process. It will happen at different times and in different ways for each child.

Advice for parents

Make your family a ‘safe space’ where your child can ask questions about achondroplasia and express themselves without fear or judgement. Answer your child’s questions with sensitive, age-appropriate answers.

Encourage your child to see their ‘differences’ as traits that make them unique and diverse. Treat your child with love and respect, and encourage healthy ideas about achondroplasia.

If you haven’t started doing so already, you might want to begin teaching your child strategies to help deal with physical or social challenges they may face. For example, if another child says something hurtful.

There are children’s achondroplasia booklets that might help your child learn about the condition as their self-awareness develops. These are available online or you can contact a local achondroplasia patient association to find good examples in your language.

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Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

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8

Child starts to develop self-awareness

Some parents find it emotionally challenging when their child begins to develop awareness of achondroplasia.[9,12] Speak with your healthcare team or contact an achondroplasia patient association about finding mental health support if you or your child would find this helpful.

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

Child starts to develop self-awareness

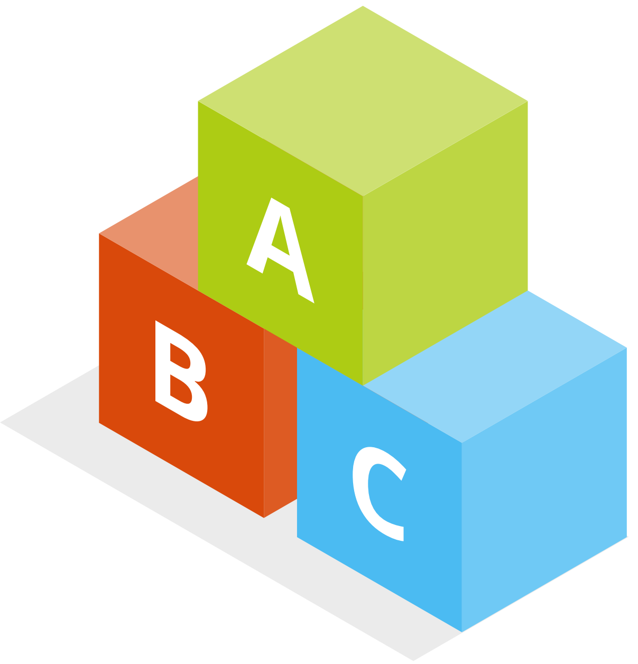
Speaking about achondroplasia within your family

Normalize speaking about achondroplasia in your family. You may want to talk with your child, and any of their siblings, about diversity. Explain that everybody is different (e.g., different shapes and sizes, with different eye, hair and skin colours), but deserves the same level of respect.

Helping your child talk to others about achondroplasia

Depending on your child's age, you could teach them ways to speak to others about their achondroplasia:

- Help them develop a simple explanation of achondroplasia so they are able to tell people about it in their own words.
- It may be helpful to teach your child how to gently correct someone who uses wrong or discriminatory words.



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Birth to age 2
Ages 3 to 6
Ages 7 to 12
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9

Child starts nursery or preschool

You
Healthcare
Social

What can parents expect at this stage?

Starting nursery or preschool is an important time for most children. Knowing what to expect, and how you can work with teaching staff to help your child settle, will help you feel more comfortable with being apart for longer periods of time.

Your child does not need to attend a ‘special’ nursery or preschool because they have achondroplasia. You should choose a nursery or preschool as you would for any other child.[23]

Advice for parents

Visit potential nurseries or preschools to check if their facilities will be suitable and speak with staff about adaptations your child may need. Most early childhood providers will be happy to meet specific adaptations to help your child be as independent as possible.[22]

Make sure nursery or preschool staff are aware of your child’s limitations in terms of motor skills. For example, they may struggle to hold pens, pencils or crayons.[22,24] This can make it harder to write, draw, and join in with arts and crafts. Staff should work with you to adapt to your child’s needs. For example:

- Using adapted materials.
- Allowing more time to complete tasks.

There may be a delay in self-care skills, so support may be needed to help your child:

- Dress and undress.
- Wash their hands.
- Use the toilet.

Be prepared to work with their nursery or preschool to make sure that they understand achondroplasia. You could share information from anchondroplasia patient associations and this Roadmap.

Some nurseries/preschools will offer individualized education or support plans, and some have dedicated members of staff, such as special educational needs coordinators.[25]

Regularly check in with staff to see how your child is managing and encourage them to help your child join in with their peers. You can give the nursery or preschool regular feedback to make sure your child continues to get the right support.

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Ages 7 to 12
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Child starts nursery or preschool

Before birth (pre-natal)
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Ages 3 to 6
Ages 7 to 12
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Healthcare considerations

Occupational therapists can suggest adaptations or tools that might help your child in a nursery or preschool environment.[21,22] For example, ways to make writing easier if your child has reduced hand movement. Speak with your healthcare team about meeting an occupational therapist if you need advice.

Your child will continue to be monitored by their healthcare team on a regular basis. You may want speak with their nursery or preschool about how this will impact attendance.

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9

Child starts nursery or preschool

Speaking to nursery and preschool staff about achondroplasia

Meet with your child's teachers before they start nursery or preschool to talk with them about achondroplasia and answer any questions.
You can explain:

- What achondroplasia is.
- What it might mean for your child (for example, they may need additional support with activities like writing and toileting).
- Signs of common health problems they can look out for.

Reasonable adaptations can then be made. Any problems can be addressed as soon as they happen.

Achondroplasia patient associations have resources that can be helpful when speaking with schools, nurseries, preschools, and teaching staff.

Preparing staff to manage social concerns

Talking to staff about achondroplasia can help them manage the way other children react to and interact with your child.

Some preschool children may be completely unaware of any differences in their classmates. Other children may ask questions or make hurtful comments, even without meaning to. You may also face hurtful comments from other parents.

You may want to:

- Think about what you will do or say in such a situation.
- Ask staff what the policy is for handling potential bullying incidents.
- Teach your child that it is not okay to be teased or bullied. Talk to staff as soon as possible if this happens.

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
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10 Child starts to make friends

You
Healthcare
Social

What can parents expect at this stage?

Your child will begin to form friendships with other children. Sometimes, your child may find it difficult to join in with certain activities or games.

Advice for parents

Support your child in meeting other children and making friends, as this will build their self-confidence.

Help your child find activities they enjoy. For example, team sports are a great way to meet children with similar interests and help your child form friendships. Let the adult running the activity know if any adaptations or equipment might be needed for your child to take part.

If you can, give your child space to explore social opportunities. Letting go can be difficult, but it can also be rewarding to watch your child make their own way and begin making friends with other children.

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
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10 Child starts to make friends

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

Before your child begins a new sport or activity, speak with their healthcare team to check that it's safe.[26] Discuss how your child might be able to join in.

Questions to ask your healthcare team

- Are there any sports or activities that my child should avoid?
- What adaptations or equipment are available to help my child play certain sports or activities?

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10

Child starts to make friends

Ages 3 to 6

You

Healthcare

Social

Speaking to your child’s friends and their parents about achondroplasia

Your child’s friends or their parents may ask questions about achondroplasia. You may want to:

- Prepare a simple explanation, describing it as part of human diversity, rather than a difference or a problem.
- Use the same language in your explanations with different people.
- Let friends’ parents know of any help or adaptations your child needs. This way, they will feel more comfortable being responsible for them during a play date, for example.
- Teach your child that it is not okay to be teased or bullied. Talk to the responsible adult as soon as possible if this happens.

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
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Child starts primary school

7. Child becomes independent of parents
8. Child starts to develop self-awareness
9. Child starts nursery or preschool
10. Child starts to make friends
11. Child starts primary school

What can parents expect at this stage?

Your child will usually begin primary school between 4 and 6 years old. Your child does not need to attend a 'special' school because they have achondroplasia.[23]

When choosing a school, consider the same factors you would for other children, as well as the school's layout and buildings.[23] For example, lots of steps might be physically difficult for your child.

Advice for parents

Visit potential primary schools before your child starts, to check the facilities are suitable. Speak with teaching staff about possible adaptations your child might need.

You may be asked to meet with the school's special educational needs coordinator, or you might request this yourself. These are specialist staff members who can offer individualized education plans and organize things to help your child in school.

Speak with your child about how they are feeling about starting school and reassure them of what they have to look forward to.

Accessibility

It is helpful to go with your child to clarify any doubts or concerns you may have with the school building and related facilities. Things to look out for include:[27]

- Are there accessibility buttons to open doors (a permanent step may be needed)?
- How far away is the drop-off area from their classroom?
- How will your child get to school? Is there a school bus in your local area?
- Can your child reach their locker or coat hook?
- Can your child sit comfortably at desks/on the chairs provided?
- Can your child access bathroom doors, toilets, and drinking fountains?
- Can your child use the canteen/lunchroom counters?

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Child starts primary school (cont.)

You may consider speaking with the school about possible adaptations that have helped your child before, such as:[21,22]

- Writing aids, like jumbo pencils.
- Using a tablet/laptop for written work, or wide-lined notebooks.
- Allowing more time for moving between classrooms.
- Steps to help reach the blackboard or to give foot/leg support while seated.
- Steps or a stool for toilet and sink access.
- Customized chairs or cushions for chair backs.
- A lower locker.
- Extenders for reaching handles or light switches, or to help when going to the toilet.

Self-care

Let the school know if your child is struggling to manage self-care skills, for example, toileting. At the same time, encourage staff to let your child be as independent as possible. This will help them build their self-confidence in their new environment.

Physical education

Some children with achondroplasia find exercise particularly tiring (known as ‘exercise intolerance’). This may be due to comparatively low muscle strength, needing to move their bodies in certain ways, or tiredness.[16] This might impact how your child takes part in physical education. You may want to:[23]

- Inform teachers of any issues to be aware of, such as avoiding pressure on the back or joints, or impact activities (including ‘forward rolls’).
- Advise your child to avoid rough games that involve lots of physical contact.

Ages 3 to 6

You

Healthcare

Social

Before birth (pre-natal)
Birth to age 2
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Child starts primary school

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

Your child will continue to be regularly monitored by their healthcare team. It is a good idea to let their school know how this might impact their schedule.

Occupational therapists can help suggest adaptations or tools that might help your child in a primary school environment. Speak with your healthcare team about meeting an occupational therapist if you need advice.

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11

Child starts primary school

Speaking to teaching staff / other parents about achondroplasia

Meet with teaching staff before your child starts school to talk with them about achondroplasia and answer any questions. This can help make sure the adults around your child are well informed and are able to explain achondroplasia to the other children. This can be helpful if your child wants to visit a friend's house.

You can explain:

- What achondroplasia is.
- What it might mean for your child (for example, they may need additional support with activities like writing and toileting).
- Signs of common health problems they can look out for.

Reasonable adaptations can then be made. Any problems can be addressed as soon as they happen.

Achondroplasia patient associations have resources that can be given to schools to help explain achondroplasia and how it might impact your child's academic, personal, and social development.

Preparing teaching staff to manage social concerns

Talking to staff about achondroplasia can help them manage the way other children might react to and interact with your child.[23]

Your child may face hurtful comments from others, which can make them feel self-conscious or embarrassed. These comments or questions may not always have a hurtful intention but can still be hard to hear.[23]

You may want to:

- Ask staff what the policy is for handling potential bullying incidents.
- Think about what you will do or say in such a situation.
- Teach your child that it is not okay to be teased or bullied. Talk to teaching staff as soon as possible if your child is being bullied.

Before birth (pre-natal)
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Ages 7 to 12
Ages 13 to 18
Healthcare considerations

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12

Child gains more awareness of achondroplasia

Ages 7 to 12

You

Healthcare

Social

12. Child gains more awareness of achondroplasia
13. Child starts secondary school

What can parents expect at this stage?

As your child gets older, they will begin doing things on their own and developing relationships with a wider social circle. They may become more aware of their achondroplasia and start to see themselves as different from their peers.

Older children may notice differences between themselves and their peers as they enter puberty, when physical differences become more obvious.

This growing awareness of achondroplasia and its impact on self-image can be difficult for children and their families.[28] Your child may feel distant from their peers and blame their condition.

Advice for parents

You should be prepared to answer questions about your child's self-image and identity. If your child is entering puberty, talk about what this will mean for them.

Use positive messaging about achondroplasia and the fact that your child's worth is not measured by their physical size. Many achondroplasia patient associations have resources that can explain achondroplasia to children and boost their self-image.

Dress your child in age-appropriate clothing, even if this means making alterations so they fit comfortably. This will help build your child's self-esteem and encourage people to treat them according to their age rather than their size.

Your child may feel like withdrawing from leisure activities. Encourage them to keep going. Speak with the responsible adults about how your child is feeling.

Achondroplasia patient associations may be able to provide opportunities for your child to socialize with other children with achondroplasia. Encourage your child to find friends with or without achondroplasia.

Before birth (pre-natal)
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12

Child gains more awareness of achondroplasia

You
Healthcare
Social

As your child gets older, they may start to become more worried about check-ups and treatments.[6] Try to reassure your child that you are there to support them throughout.

It may be a good time to let your child get more involved in conversations with their healthcare team. Encourage them to ask about anything they are unsure of or don't understand. For example:

- Why they have achondroplasia.
- Why they are experiencing certain things related to their health.

Continue to encourage regular exercise and a healthy diet to help your child maintain a healthy weight.[16] Increased body weight will have an impact on your child's joints, affecting their mobility and independence.[16] Regular exercise, meanwhile, will help their overall health and mental wellbeing.

Questions to ask your healthcare team:

- Can we involve my child in our conversations about their achondroplasia?
- Would you be happy to answer questions my child has about achondroplasia?

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Child gains more awareness of achondroplasia

Helping your child respond to questions about achondroplasia

Your child may start to notice comments or reactions from people around them about their achondroplasia. Speak with your child about how they could respond in these situations. For example:

- Walking away.
- Ignoring the comment.
- Providing simple answers or explanations about achondroplasia.
- Speak to a trusted adult, such as a teacher.

Talking about achondroplasia within your family

Normalize speaking about achondroplasia in your family so your child, and any siblings, know that it is nothing to be ashamed or embarrassed about. Make your family a ‘safe space’ for your child, where they can share their feelings and ask questions without judgement.

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Child starts secondary school

You

Healthcare

Social

What can patients expect at this stage?

Entering secondary education is an important time for your child's personal, social and educational development. Special schools are not needed for children with achondroplasia.

Entering secondary education means taking increased personal responsibility for their education and interpersonal relationships. You can support your child to face this period of change with confidence.

Advice for parents

You may want to:[21,22,27]

- Visit potential secondary schools to assess the layout and environment.
- Discuss possible adaptations with teaching staff.
- Be prepared to explain achondroplasia and what it means for your child's academic and physical capabilities.
- Speak with the school's special educational needs coordinator about an individualized plan to support your child.
- Involve your child in decisions about which adaptations or equipment they need and want to use.

It might be possible to carry through adaptations that worked for your child in primary school to secondary school. For example, using a step stool. Before your child starts school, think about what has worked before.[23]

Adaptations that might be introduced at secondary school include:[22,27]

- Lightweight folders, to avoid needing to carry too many books.
- Having 2 copies of important textbooks or workbooks, 1 for home and 1 for school, to avoid the need to carry books back and forth.
- Increased use of digital media and tools in teaching methods and in completing schoolwork.

Depending on where you live, your child's school may be able to apply for funding for any particular equipment, if needed. Contact your local achondroplasia patient association for more information.

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13

Child starts secondary school

Ages 7 to 12

You

Healthcare

Social

12. Child gains more awareness of achondroplasia
13. Child starts secondary school

Your child will continue to be monitored by their healthcare team on a regular basis. It is a good idea to speak with their school about the possible impact on their schedule.

You may choose to speak to the school to make sure their education isn't greatly impacted:[27]

- Ask if classwork can be provided in advance to be completed at home.
- Ask if deadlines can be extended as needed.
- Ask if tests or exams can be taken on different days to other children.

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Child starts secondary school

Speaking to teaching staff about achondroplasia

Meet with teaching staff before your child starts secondary school to talk with them about achondroplasia and answer any questions.

You can:

- Explain achondroplasia to your child's teachers if you feel comfortable doing so.
- Mention signs of common health problems they can look out for.
- Make sure your child is included in peer activities and let the school know of any adaptations needed.

Some children with achondroplasia face difficulties around inclusion in school. Sometimes this is due to school staff separating them from their peers, often for physical reasons. This can impact self-esteem and school performance.[28]

Achondroplasia patient associations have resources that can be given to schools to help explain achondroplasia and how it might impact your child's academic, personal, and social development. Some associations also host meetings with school staff and parents to explain the impact of achondroplasia on a child's school experience.

Preparing teaching staff to manage social concerns

Talking to staff about achondroplasia can help them manage the way other children might react to and interact with your child. You may want to ask staff what the policy is for handling potential bullying incidents.[23]

Teachers should be role models for how your child should be treated. You can:

- Let your child's teachers know what type of language to look out for that could be hurtful.
- Tell them what words you prefer to use when talking about achondroplasia.

Before birth (pre-natal)
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14

Young person’s sense of self-awareness develops further

Ages 13 to 18

You

Healthcare

Social

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
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Ages 13 to 18
Healthcare considerations

What can parents expect at this stage?

As your child matures, their condition will become more physically apparent. Their sense of self-awareness – and awareness of their achondroplasia – will continue to develop. They may also become more aware of social issues that can affect people with achondroplasia.[29,30]

Sometimes, people with achondroplasia face discrimination, impacting things like self-esteem, education and employment. This is often caused by cultural stereotypes or incorrect beliefs about people with achondroplasia – linked to the media and advertising, movies, stories, or even mythologies.[29,30]

It is important to teach your child to be confident and to advocate for themselves. The more confident your child feels in themself, their bodies, and their place in the world, the more equipped they will be for adult life.

Advice for parents

Teach your child that their success and place in society are not linked with their physical height. It is their character and abilities that will determine their future, including how they see themselves. Always speak positively about their future options and encourage them to aspire to achieve their goals.

You can help build your child’s self-esteem by:

- Encouraging ‘body positivity’ through praise about their appearance and behavior.
- Praising your child when they overcome an obstacle or achieve something.

Teach your child that it is never acceptable to be bullied, or receive insulting or discriminatory remarks.

Look out for signs your child may be struggling, such as becoming more withdrawn or refusing to take part in activities they used to enjoy. Encourage them to keep taking part in social activities and to be resilient.[31]

It can be helpful for people with achondroplasia to spend time with others with the condition or to join online communities. Look for achondroplasia patient associations that offer opportunities to meet other people with achondroplasia, in person or in online groups.

Young person's sense of self-awareness develops further

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

Concerns about self-image can impact a person's mental health and, in some cases, may lead to conditions such as depression.[31]

Depression is more common in people with achondroplasia than the general population, so it is a good idea to be aware of the signs. These include:[31]

- An ongoing sad, anxious, or 'empty' mood
- Feelings of hopelessness
- Irritability, frustration, or restlessness
- Loss of interest in hobbies
- Decreased energy
- Difficulty sleeping

If you see these signs or have any worries about your child's mental health, contact your child's healthcare team or a mental health professional straight away. For general mental health support and awareness, contact your local achondroplasia patient association for advice.

Questions to ask your healthcare team

- What can I do to help my child's mental wellbeing?
- What mental health support is available for my child if they need it?
- Who will be involved in providing mental health care?

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14 Young person's sense of self-awareness develops further

Teaching your child to address discriminatory behaviour

Sometimes, your child may want to speak out against discriminatory behaviour in public.

You can:

- Help your child plan and practice what they might say or how they would respond if this happens to them. You might have your own experiences you can share with them about how you handled similar situations.
- Speak with your child about how they would deal with any situations at school and their options are for raising issues with teachers.

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Healthcare considerations

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15

Young person begins to seek teenage independence

You
Healthcare
Social

What can parents expect at this stage?

During their teenage years, your child will become more independent. They may start socializing more with friends. They may start to transport themselves around, either by public transport or in their own car.

Everyone with achondroplasia matures at a different rate. Look out for signs your child wants or needs more freedom. Be ready to encourage this when it does happen.

People with achondroplasia can learn to drive to gain independent mobility. To safely drive a car, people with achondroplasia will need a vehicle with adaptations. For example:

- pedal extensions.
- raised seat cushions.
- switch off their airbag (rules vary by country).

Entering the workplace is another milestone for many teenagers. This could be full-time employment or a casual job. There may be some professions that people with achondroplasia find difficult to access. For example, some trades that require a lot of physical mobility.

Advice for parents

Aim to strike a balance between offering support and encouraging independent activities , if they fit with your parenting style and family boundaries.[32] For example, you could encourage your teen to buy their own clothes, even if they need adjusting.

If you have an older teen who is reluctant to do things independently, speak with them about how they are feeling. Encourage independence at the rate that is appropriate and comfortable for your child.

Encourage your teen to help with tasks around the home. This might include house cleaning or laundry, where adaptations might be needed to help with vacuuming, reaching surfaces, accessing top-loader washing machines, etc.

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Young person begins to seek teenage independence (cont.)

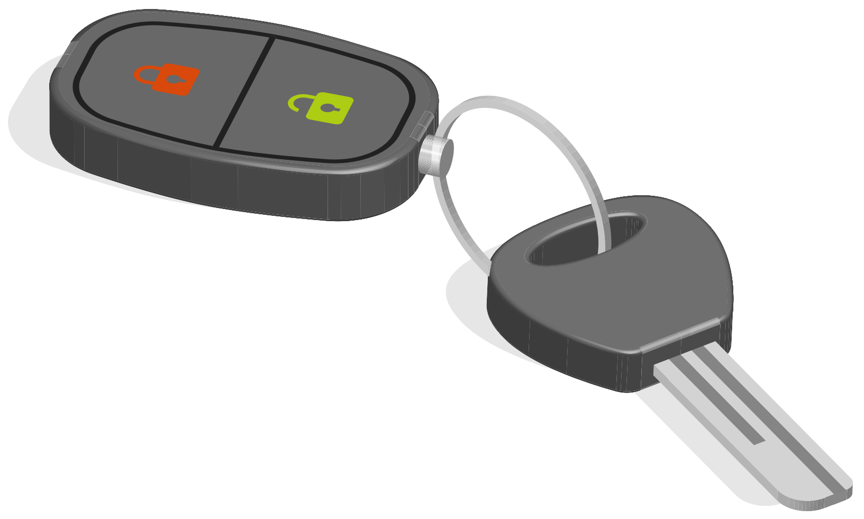
Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

You can help your child with learning to drive by:[33]

- Looking for driving instructors or driving schools that have experience teaching people who need adaptations to drive.
- Discussing specific needs and adaptations with the chosen instructor/school.
- Researching adaptations and where to find them with achondroplasia patient associations.
- Researching how your insurance might be affected or what insurance options there are for your child
- Researching the legal background where you live, for example, regarding airbag on-off switches or other adaptations

You can support your teen to find work by:[32]

- Identifying their strengths and abilities rather than limitations.
- Arranging 'work experience' periods at appropriate workplaces to improve their confidence and experience of the workplace.
- Finding employment advice from qualified advisors.
- Researching anti-discrimination laws relating to workplace hiring, just in case.
- Contacting achondroplasia patient associations for advice.



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15 Young person begins to seek teenage independence

Throughout their teenage years, your child's regular healthcare check-ups will continue, including regular checks for:[14]

- Sleep apnea and hearing issues.
- Possible orthodontic issues.
- Pain and fatigue.

As your child prepares to transition to adult care, they should become more involved in conversations with their healthcare team.[32] Ideally, you should aim to play a minimal role in these conversations for older teens. However, be prepared to speak up on behalf of your child or ask questions if needed.

Speak to your teenager before check-ups to see how much involvement they want you to have, and in the case of older teens whether they would like you in the doctor's office with them or to wait outside.

Encourage them to ask questions and try not to speak on their behalf (unless you need to). This will get them used to speaking with doctors and understanding important information.

Before birth (pre-natal)
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Young person begins to seek teenage independence

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

Preparing your child to support themselves within society

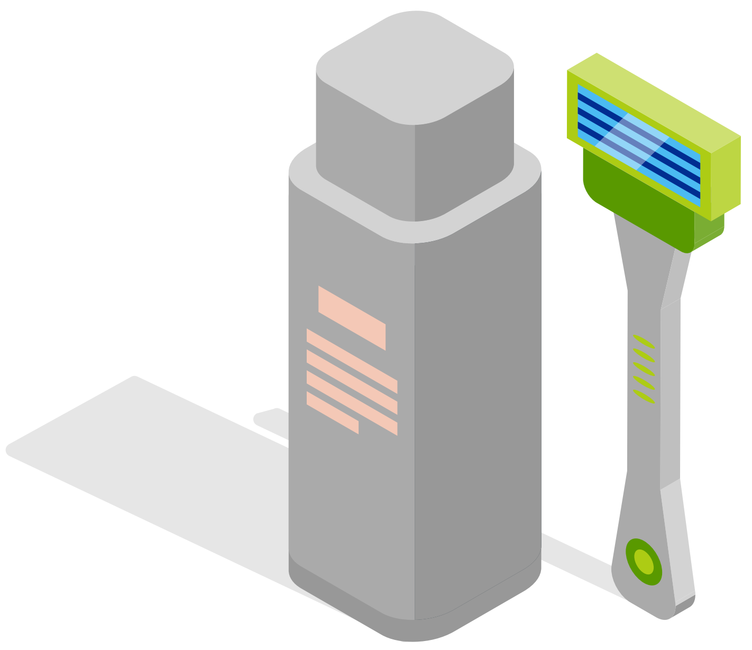
As your teen learns to live independently, it is important they can be proactive in finding help or advice, as well as in advocating for themselves. For example, during the process of finding work.

Along with your teen, you can:

- Plan and practice what they might say or do if they hear discriminatory language, for example, during a job interview.
- Remind your teen how they could respond in these situations – ignoring the comment, or providing simple answers or explanations about achondroplasia.

You and your teen probably already have a standard way you describe achondroplasia to people around you. It might be a good time to review this description and the wording you prefer people to use to talk about achondroplasia.

If you haven't done so already, you can introduce your child to achondroplasia patient associations so that they know these are valuable sources of support and information.



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Sex and relationships

What can parents expect at this stage?

At some point, your teen will probably have questions about sex and relationships. Understanding what questions or concerns they may have will allow you to have an open and honest conversation when the time is right.[34]

Young people with achondroplasia will sexually develop in the same way as their peers.[1] However, people living with conditions such as achondroplasia can be dismissed as non-sexual beings by the general population.[35] This can negatively impact their self-confidence and ability to find a partner.[36]

There will be different approaches to this topic depending on your family's cultural background, beliefs, and preferences. The below is advice only: it is important to do what is right for you and your family.

Advice for parents

Let your child know that they will feel the same feelings and experience the same physical changes during puberty as their peers.[36]

Reassure them that:[36]

- People with achondroplasia have the same rights as everyone else when it comes to having a partner, if this is something they would like.
- There are many people with achondroplasia in loving, long-term relationships.
- Their attractiveness to a partner is not limited by their physical height.

Be prepared to discuss intimacy with your teenager. Young people with achondroplasia may need to find ways to be intimate that overcome physical limitations they or their partner may have.[14,34] These will be different for couples where both partners have achondroplasia compared with those where one partner has achondroplasia and the other is average height.

Advise your teenager what to do if they encounter coercive or predatory sexual behaviour.[29]

There is little official advice about this topic, but achondroplasia patient associations will be able to offer advice if you or your teenager contact them.

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Sex and relationships

Ages 13 to 18

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14. Young person's sense of self-awareness develops further
15. Young person begins to seek teenage independence
16. Sex and relationships
17. Transition to adult healthcare

Your child's fertility will not be affected by achondroplasia. An important topic to discuss is the genetic risk of them having a child with achondroplasia. This risk will change depending on their partner, with the risk increasing if both partners have achondroplasia.[14] Your child may wish to discuss this with a healthcare professional.

You may have had some advice on this if you had genetic counseling in the past. Now, it is important for your child to have this information as well. This will inform their decision-making around intimacy and family planning.

Speak with your healthcare team if you need information about genetic risks or about access to genetic counseling. If you prefer, ask your healthcare team if they are able to give intimacy and contraception advice directly to your teenager.

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Sex and relationships

Preparing your teenager to talk about achondroplasia with partners

You may want to speak to your teenager about how they will discuss achondroplasia with potential future partners.

You could support your teen by:

- Thinking together about the wording they would prefer partners to use to talk about achondroplasia.
- Planning and practicing what they might say or do if they hear discriminatory language in the context of an intimate relationship.
- Exploring how they will speak to potential future partners about any physical difficulties they have that could impact intimacy.

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Transition to adult healthcare

Ages 13 to 18

You

Healthcare

Social

What can parents expect at this stage?

Between the ages of 16 and 18, your child will transition to the adult healthcare system.[37] They may need your support managing this transition.

Your child will become more responsible for decision-making about their own healthcare. They may do things you do not agree with, like missing appointments or deciding against treatments.[38] They may prefer to do other activities or socialize instead.

Try to allow your teen a certain level of freedom to manage their own healthcare. During this time of change, continued emotional support from friends and families can help your child not feel too disoriented.

Depending on where you live, health insurance coverage might change as your child becomes an adult, so you may need to prepare for this.[37]

Advice for parents

You can empower your child to manage their own care, but you may also need to provide some help. For example, you may want to:

- Emphasize how important regular follow-ups are.
- Teach or show your child how to communicate their needs to healthcare professionals.
- Support with practical things like refilling prescriptions, booking or travelling to appointments.

Discuss any concerns with your child. The transition to a new healthcare team may be difficult for you both.[37,39–41]
You might have built a close relationship with your current healthcare team.

If you haven't already, introduce achondroplasia patient associations to your child. They:

- Are a source of additional support and information.
- May be able to help with the administrative transition process.
- Could put you in touch with people who have been through this transition.

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Transition to adult healthcare

Ages 13 to 18
You
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Social

There is no 'one size fits all' approach or a definite 'right' time for your child to transfer to adult healthcare services – although it is usually between 16 and 18 years old.[37]

Start having conversations with your child and healthcare team about the transition as early you can, to make sure the process is as smooth as possible. Your current team may help create a transition plan and advise on specialists with experience treating achondroplasia.

In adulthood, care will focus on maintaining health and ability to go about their daily life, with as few pain or mobility issues as possible. People with achondroplasia:[6,37,42]

- May continue to face some of the health issues they had in childhood, such as sleep apnea and spine issues.
- May develop hearing issues earlier than other people, so routine screening may be necessary.
- Are at a higher risk of obesity, so weight management is important.
- May experience ongoing pain in their back, joints, and legs, which can impact their well-being and mood.

It is important your teen is aware of and has access to their own medical history.[37] You or your child should ask your healthcare team to provide this when entering adult care.

Questions to ask your healthcare team

- How will adult healthcare services differ?
- Do you have a standard care transition plan?
- How often will my teen have to attend appointments once they transition to adult care?
- How do you make sure my teen's medical history is handed over so that they do not experience a drop in their standard of care?

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Transition to adult healthcare

Ages 13 to 18

You

Healthcare

Social

Preparing your child to have discussions with healthcare professionals

As they transition to adult care, your child – now a young adult – will need be their own best advocate and lead discussions about their health. You can help to prepare them for these conversations.

It is important to know that the adult care system does not provide as much support for people with achondroplasia as the child care system, so preparing them to advocate for themselves will be particularly important.[37,39–41]

As healthcare services become less specialized in achondroplasia, they may experience stigma in these settings.[29] You may talk to your teen about what they might say or do if they hear discriminatory language in a healthcare setting.

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Between birth and age 2

Healthcare considerations

You

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Social

Your child will be closely checked and monitored by their healthcare team during early childhood.[6,10,14,24,43,44] This is to track their developmental milestones and look out for physical problems that can impact children with achondroplasia. Feel free to ask your doctor questions about what they see on the scans and tests for your understanding.

Your healthcare team may recommend:

- Scans of the brain, neck, and spine, to check for spinal issues:[6]
  - These will check for an issue called foramen magnum stenosis, where the spinal cord, one of the main nerves in the body, is compressed by the bones of the head. Regular scans are a reliable way of tracking this issue.
  - Your child may have at least one MRI in the first year of life. Additional MRIs may be needed.
- A sleep study to detect sleep apnea:
  - If your child is showing signs of sleep apnea (when breathing stops and starts during sleep), their doctor may recommend a sleep study to check how this is affecting their health and to see what is causing it.[1,6,45] A sleep study will be recommended before your child turns 1 year old.[6,10]
  - Signs to look out for yourself when your baby is asleep include: breathing that starts and stops during sleep, loud snoring, gasping for air during sleep and changes in skin colour.[45]
- A hearing test, to look for signs of middle ear infection:
  - Hearing tests can help doctors see if your child has trouble hearing, which might be a sign of an ear infection.[46] This is common and treatable in children with achondroplasia.[6,10]
  - Signs to look out for include: tugging or pulling at the ear(s), fussiness and crying, trouble sleeping, fever, fluid draining from the ear, trouble hearing or responding to quiet sounds.[46]
- Meeting with an ear, nose and throat specialist to evaluate potential concerns related to position of the tongue, low muscle tone (hypotonia), and breathing.[6]

Your child might experience other rare complications associated with achondroplasia, which will need further tests. More information about these complications can be found by visiting achondroplasia patient association websites.

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Between birth and age 2 (cont.)

Healthcare considerations

You

Healthcare

Social

Before birth (pre-natal)
Birth to age 2
Ages 3 to 6
Ages 7 to 12
Ages 13 to 18
Healthcare considerations

As with any child, speak with a healthcare professional if you feel worried about their health. There are no silly questions, and you won't be wasting anyone's time.

Treatments and therapies

You may hear about certain treatments that are designed to help physical development in a child with achondroplasia. It is a good idea to research these options and discuss them with your healthcare team.

Surgical treatments

- If scans of your child's brain, head, and neck show spinal issues, your healthcare team may suggest surgical treatment. A common surgery for children with achondroplasia is a procedure to reduce pressure on the spinal cord if it is being compressed by the bones of head and neck (foramen magnum stenosis).[6] There are also things you can do as parents to help prevent spinal issues.
- Other surgical options are available for children with specific issues, when suggested by specialists.

Treatment for middle ear infections

- Ear infections are common in children with achondroplasia.[10] Some children who get repeat infections might need ear tubes put in, which can help lower the chances of further infections.[20]

- Ear tubes will be put in during an operation. Your child will be given a general anesthetic, so will be unconscious and pain-free during the procedure. Older children and adults don't always need to be fully asleep for this treatment, so make sure you check what will happen with your child.[20]

Treatment for sleep apnea

- Some children with sleep apnea need an operation to remove adenoids. Adenoids are small areas of fleshy tissue in the back of the throat that can make breathing difficult for some people.[6]

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Between birth and age 2 (cont.)

You

Healthcare

Social

You child may experience some pain after different treatments – although this is normal, you should discuss pain relief options with their healthcare team. Make sure your home is ready for your child when they return home, so everything they need is close by and things like transport, clothing, and furniture are arranged.

Certain therapies may be also available to your child. For example, osteopathy or aqua therapy, which can improve the quality of life for children with achondroplasia.[10]

You may want to explain tests and procedures to any other children in the family so they are also aware of what will happen to their sibling with achondroplasia. Help them understand that their sibling might need to visit hospitals regularly and that this might impact their routines if a parent has to be away.

Before birth (pre-natal)
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Between ages 3 and 6

Healthcare considerations

You

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Social

18. Between birth and age 2
19. Between ages 3 and 6
20. Between ages 7 and 12
21. Between ages 13 and 18

Your child will be regularly checked by their healthcare team to look for problems that can affect them between the ages of 3 and 6 years.[6,14] Their healthcare team will also continue to track developmental milestones, like walking and speech development. Make sure you attend these check-ups so that any such issues can be found as early as possible. Continue to contact achondroplasia patient associations or speak with your healthcare team for any advice you might need.

Treatments and therapies

If you notice anything you're worried about, seems unusual for your child, or is causing obvious problems, speak with your healthcare team. They will work with you to make a decision on what treatment or support, if any, your child needs.

During this time, your child may need the following:

- Treatment for middle ear infections
  - Please refer to information on treatment of ear infections in the 'Between birth and age 2' section of the roadmap
- Treatment for sleep apnea
  - Please refer to information on treatment for sleep apnea in the 'Between birth and age 2' section of the roadmap.
- Treatment for teeth and dental problems
  - Children with achondroplasia may have dental problems, such as teeth being 'crowded'. When your child is around 5–6 years old, they should be referred for a check-up with a specialist dentist called an orthodontist, to see if any treatments are needed (e.g., braces or a palatal expansion, a device fitted to widen the upper jaw).[6,47]
- Help with speech
  - Some children have ongoing issues with speech or communication, in which case your child may be seen by a specialist in speech and language therapy.[6]

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Before birth (pre-natal)
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Between ages 3 and 6 (cont.)

- Support for problems with movement and posture
  - If your child has ongoing problems with movement or posture, they may be referred to a physiotherapist. For severe issues with the legs, arms, back/neck, or other joints, your child may need to be checked by an orthopedist.[6]
  - Between the ages of 2 and 12 years old, some children with achondroplasia develop bowed legs, or this can become more pronounced. Ask your healthcare team for advice if this is something you have concerns or questions about.[6]

You child may experience some pain after different treatments – although this is normal, you should discuss pain relief options with their healthcare team. Make sure your home is ready for your child when they return home from treatments, so everything that they need is close by and things like transport, clothing, and furniture are arranged.

Your child may continue some of the regular therapies he/she has been receiving since early childhood. For example, osteopathy, speech therapy or aqua therapy, which can all improve the quality of life for young people with achondroplasia.

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Social

Before birth (pre-natal)
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Between ages 7 and 12

Healthcare considerations

You

Healthcare

Social

Your child will be regularly checked by their healthcare team to look for problems that can affect them between the ages of 7 and 11 years.[6,14] Make sure you attend these check-ups so that any such issues can be found as early as possible. Continue to contact achondroplasia patient associations or speak with your healthcare team for any advice you might need.

Treatments and therapies

If you notice anything you're worried about, seems unusual for your child, or is causing obvious problems, speak with your healthcare team. They will work with you to make a decision on what treatment or support, if any, your child needs.

During this time, your child may need the following:

- Treatment for middle ear infections
  - Please refer to information on treatment of ear infections in the 'Between birth and age 2' section of the roadmap
- Treatment for sleep apnea
  - Please refer to information on treatment of sleep apnea in the 'Between birth and age 2' section of the roadmap
- Treatment for teeth and dental problems
  - Before 6 years old, your child should have been referred for a check-up with a specialist dentist called an orthodontist. It is common for children with achondroplasia to have dental problems, such as teeth being 'crowded' or having an 'underbite'.[6,48]
  - If your child needs dental surgery, they will be examined to confirm the safest option for pain relief.[6,48]
  - Your child will have routine check-ups with general dentists throughout their childhood and teenage years. The dentist will give advice on good dental habits, such as diet advice, cleaning techniques, and products, and monitor the growth patterns of your child's teeth and jaw.[6,48]
- Help with speech
  - Some children have ongoing issues with speech or communication, in which case your child may be seen by a specialist in speech and language therapy.[6]

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Before birth (pre-natal)
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Between ages 7 and 12 (cont.)

- Problems with movement and posture
  - If your child has ongoing problems with movement or posture, they may be referred to a physiotherapist. For severe issues with the legs, arms, back/neck, or other joints, your child may need to be checked by an orthopedist.[6]
  - Between the ages of 2 and 12 years old, some children with achondroplasia develop bowed legs, or this can become more pronounced. Ask your healthcare team for advice if this is something you have concerns or questions about.[6]

Treatments and therapies

You may hear about certain treatments that are designed to help a child with achondroplasia. It is a good idea to research these treatments and discuss them with your healthcare team.

Your child may experience some pain after different treatments – although this is normal, you should discuss pain relief options with their healthcare team. Make sure your home is ready for your child when they return home from treatments, so everything that they need is close by and things like transport, clothing, and furniture are arranged.

Your child may continue some of the regular therapies he/she has been receiving since early childhood. For example, osteopathy, speech therapy or aqua therapy, which can all improve the quality of life for young people with achondroplasia.

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Between ages 13 and 18

Healthcare considerations

You

Healthcare

Social

Your child will be regularly checked by their healthcare team to look for problems that can affect them between the ages of 13 to 18 years.[6,14] Make sure you attend these check-ups so that any such issues can be found as early as possible. Continue to contact achondroplasia patient associations or speak with your healthcare team for any advice you might need.

During their teenage years, encouraging an increasing amount of independence and responsibility for their own healthcare will help your child transition into adult care when they reach adulthood.

Treatments and therapies

In later teenage years and into young adulthood, joint, leg, and back pain can be common. If your child has problems with movement or posture, or finds certain actions difficult or painful, they may be referred to a physiotherapist. For severe issues with the legs, arms, back/neck, or other joints, you child may need to be checked by an orthopaedist. Elbow stiffness may get worse with age – this will be monitored by your healthcare team.[6]

There are some specific medical issues that your healthcare team will ask about during your child's teenage years, including:[6,49]

- Their ability to do certain tasks like self-care, driving, and general daily functioning. This gives the doctors an overall sense of their wellbeing and independence. Your child might be asked if they need any adaptive equipment or mobility devices to help maximize their independence. They may be referred to an occupational therapy for advice.
- How they find managing their weight, which can have an impact on general health, joint health, and independent mobility. Your healthcare team will provide information on exercise and the importance of eating a healthy diet to maintain your child's strength, overall health, and flexibility.
- Appropriate exercise and/or sports, to minimize the risk of injury or complications. This is important at this stage because your child will be doing more activities on their own, and physical activity will help with their mood and mental health and keep their weight at a healthy level.

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Additional Resources

- Beyond Achondroplasia (English/Portuguese/Spanish/Russian)
  https://www.beyondachondroplasia.org/en/
- Beyond Achondroplasia – Achondroplasia Challenges (English/Portuguese/Spanish/Russian)
  https://www.beyondachondroplasia.org/en/health/achondroplasia-challenges
- The first European consensus on principles of management for achondroplasia (English)
  https://pubmed.ncbi.nlm.nih.gov/34332609/
- Those who live with Achondroplasia have stories that go beyond what is seen (Spanish):
  https://achondroplasia.biomarin.com/es-419/mihistoriavamasalla/
- A little Polish story book - understanding dwarfism (Polish)
  https://understandingdwarfism.com/a-little-story-book
- The roof of the world story book (Italian)
  https:/www.carthusiaedizioni.it/libri/511/il-tetto-del-mondo
- 'Nanismo' written by Velvit Severo (Portuguese)
- 'Strong and Mighty Max' (English)
  https://strongandmightymax.com/
- 'Not too big, not too small, just right for me!' (English)
  https://understandingdwarfism.com/childrens-book
- 'Let the sun shine' story book (Spanish)
  https://www.fundacionalpe.org/media/Biblioteca/Literatura_y_artes/QUE%20SALGA%20EL%20SOL.pdf
- Police short story (Spanish)
  https://www.fundacionalpe.org/media/Biblioteca/Literatura_y_artes/Cuento%20PABLO%20GARCIA%20POLICIA-%20(Garcia%20M%20y%20%20Latonda%20E).pdf
- Optimising the diagnosis and referral of achondroplasia in Europe: European Achondroplasia Forum best practice recommendations (English)
  https://pubmed.ncbi.nlm.nih.gov/35897040/
- Achondroplasia: a comprehensive clinical review (English)
  https://pubmed.ncbi.nlm.nih.gov/30606190/
- Resources covering secondary education (Spanish)
  https://www.fundacionalpe.org/es/biblioteca?categoria=educacion-y-logopedia
- Speech Therapy Practical Exercises (Spanish)
  https://www.fundacionalpe.org/media/Biblioteca/Educacion-Logopedia/praxis%20de%20logopedia2021.pdf
- Achondroplasia & Dwarfism Children's Book (English)
  https://www.achondroplasia.com/childrens-book/
- 'Little Big Sister' written by Eoin Colfer and illustrated by Celia Ivey (English)
- 'Little people: learning to see the world through my daughter's eyes' (English)
  https://littlepeoplethebook.com/
- Achondroplasia Chile Association
  https://www.acondro.cl/
- ANNABRA Brazil
  http://www.annabra.com.br/
- Associazione per l'Informazione e lo Studio dell'Acondroplasia (AISAC)
  aisac.it
- ANDO Portugal
  andoportugal.org
- Not So Little People Russia
  achondroplasia.ru
- Association des Personnes de Petite Taille (APPT)
  appt.asso.fr
- Foundation Exploring Skeletal Dysplasia Together (FEST)
  https://www.myskeletaldysplasia.org.uk/
- Fundación ALPE Acondroplasia (ALPE)
  fundacionalpe.org
- Association for Children with Achondroplasia
  odblokuj-zycie.pl

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References

1. Legare JM. Achondroplasia. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. *GeneReviews® [Internet]*. University of Washington, Seattle; 1998 Oct 12 [updated 2023 May 11].
2. Boulet S, Althuser M, Nugues F, Schaal J-P, Jouk P-S. Prenatal diagnosis of achondroplasia: new specific signs. *Prenatal Diagnosis*. 2009;29(7):697-702. https://doi.org/https://doi.org/10.1002/pd.2280.
3. Hoover-Fong J, Cheung MS, Fano V, et al. Lifetime impact of achondroplasia: Current evidence and perspectives on the natural history. *Bone*. 2021;146:115872. https://doi.org/10.1016/j.bone.2021.115872.
4. Center for Disease Control and Prevention. Genetic Counseling. Updated 2022 Sept 23. Accessed Dec 6, 2023. https://www.cdc.gov/genomics/gtesting/genetic_counseling.htm.
5. National Society of Genetic Counselors, Genetic Alliance. *Making Sense of Your Genes: A Guide to Genetic Counseling*. Genetic Alliance; 2008. https://www.ncbi.nlm.nih.gov/books/NBK115508/
6. Savarirayan R, Ireland P, Irving M, et al. International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. *Nature Reviews Endocrinology*. 2022;18(3):173-189. https://doi.org/10.1038/s41574-021-00595-x.
7. Savarirayan R, Rossiter JP, Hoover-Fong JE, et al. Best practice guidelines regarding prenatal evaluation and delivery of patients with skeletal dysplasia. *American Journal of Obstetrics & Gynecology*. 2018;219(6):545-562. https://doi.org/10.1016/j.ajog.2018.07.017.
8. ISUOG. Polyhydramnios. Updated 2019 Sept. Accessed Dec 6, 2023. https://www.isuog.org/clinical-resources/patient-information-series/patient-information-pregnancy-conditions/growth-and-fluid/polyhydramnios.html
9. Pfeiffer KM, Brod M, Smith A, et al. Assessing the impacts of having a child with achondroplasia on parent well-being. *Qual Life Res*. 2021;30(1):203-215. https://doi.org/10.1007/s11136-020-02594-3.
10. The children's hospital at Westmead. Achondroplasia (0-5 years of age): Therapy Guidelines. https://www.beyondachondroplasia.org/content/health/health_therapies/Westmead-Achondroplasia-0-5-years-of-age-Therapy-guidelines.pdf.
11. Pauli RM. Achondroplasia: a comprehensive clinical review. *Orphanet J Rare Dis*. 2019;14(1):1. https://doi.org/10.1186/s13023-018-0972-6.
12. Barriel C. Sillas, cojines, tronas y otros accesorios para ninos con adee. Accessed January 10, 2024. https://www.fundacionalpe.org/media/Biblioteca/Recomendaciones/SILLAS,%20COJINES,%20TRONAS%20Y%20OTROS%20ACCESORIOS%20PARA%20NIÑOS%20CON%20ADEE.pdf.
13. Little People of America. Guide for Raising a Child with Dwarfism. https://www.lpaonline.org/assets/documents/LPA%20New%20View%20FINAL.%20122806.pdf.
14. Hoover-Fong J, Scott CI, Jones MC, et al. Health Supervision for People With Achondroplasia. *Pediatrics*. 2020;145(6). https://doi.org/10.1542/peds.2020-1010.
15. NHS Greater Glasgow and Glyde. Information about Achondroplasia. www.nhsggc.org.uk/media/272500/achondroplasia-information-booklet_sept-22.pdf.
16. Ireland PJ, Pacey V, Zankl A, et al. Optimal management of complications associated with achondroplasia. *Appl Clin Genet*. 2014;7:117-125. https://doi.org/10.2147/tacg.S51485.
17. Beyond Achondroplasia. Speech Therapy. Accessed Dec 12, 2023. https://www.beyondachondroplasia.org/en/health/health-therapies/speech-therapy.
18. March of Dimes. Achondroplasia. https://www.marchofdimes.org/find-support/topics/planning-baby/achondroplasia#:~:text=Achondroplasia%20is%20a%20common%20cause,to%201%20in%2040%2C000%20babies.
19. Beyond Achondroplasia. Medical exams/tests. Accessed Dec 12, 2023. https://www.beyondachondroplasia.org/en/health/health-guide/medical-exams-tests.
20. WebMD. Ear Tubes. https://www.webmd.com/cold-and-flu/ear-infection/child-need-tubes-ear-infection.
21. Beyond Achondroplasia. Adaptations. Accessed Dec 12, 2023. https://www.beyondachondroplasia.org/en/resources/daily-life/adaptations.
22. Beyond Achondroplasia. School. Accessed Dec 12, 2023. https://www.beyondachondroplasia.org/en/pages/44-resources/daily-life/6-school.
23. Association restricted growth. Going to School. https://rgauk.org/wp-content/uploads/2016/09/Goingtoschool.pdf.
24. Trotter TL, Hall JG; American Academy of Pediatrics Committee on Genetics. Health supervision for children with achondroplasia. Pediatrics. 2005 Sep;116(3):771-83. https://doi.org/10.1542/peds.2005-1440
25. twinkl. Special Educational Needs Coordinator (SENCo). Accessed Dec 6, 2023. https://www.twinkl.co.uk/teaching-wiki/special-educational-needs-coordinator-senco.
26. Little People of America. School - Which Sports Are Safe For Children Born With Dwarfism? https://lpamrs.memberclicks.net/assets/documents/School.PE.Sports.pdf.

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References (cont.)

27. Wagonlander L. School Accommodations for Students with Dwarfism. https://www.lpaonline.org/assets/documents/School%20Accommodations%20LPA.ppt.
28. Pfeiffer KM, Brod M, Smith A, et al. Assessing physical symptoms, daily functioning, and well-being in children with achondroplasia. *Am J Med Genet A*. 2021;185(1):33-45. https://doi.org/10.1002/ajmg.a.61903.
29. Consensus Paper: for an EU that cares about Achondroplasia and other Skeletal Dysplasias with Dwarfism. https://www.myskeletaldysplasia.org.uk/post/eu-consensus-skeletal-dysplasias-with-dwarfism
30. Rasmussen A, Centellas DP. Achondroplasia: casting away the stigma. *Witness*. Vision Project Inc. doi: http://www.visionproject.org/images/img_magazine/pdfs/achondroplasia.pdf.
31. National Institute of Mental Health. Depression. Updated 2021. Accessed Dec 6, 2023. https://www.nimh.nih.gov/health/publications/depression.
32. Yeo M, Sawyer S. Chronic illness and disability. *Bmj*. 2005;330(7493):721-723. https://doi.org/10.1136/bmj.330.7493.721.
33. Beyond Achondroplasia. Adaptations For The Car. Accessed Dec 12, 2023. https://www.beyondachondroplasia.org/en/pages/44-resources/daily-life/76-car-adaptations.
34. Houtrow A, Elias ER, Davis BE. Promoting Healthy Sexuality for Children and Adolescents With Disabilities. *Pediatrics*. 2021;148(1). https://doi.org/10.1542/peds.2021-052043.
35. Esmail S, Darry K, Walter A, Knupp H. Attitudes and perceptions towards disability and sexuality. *Disabil Rehabil*. 2010;32(14):1148-1155. https://doi.org/10.3109/09638280903419277.
36. Zamboni B. An Exploration of Sexual Health Education Among Individuals with Skeletal Dysplasia (Dwarfism). *American Journal of Sexuality Education*. 2018;13:217-231. https://doi.org/10.1080/15546128.2018.1457463.
37. European Achondroplasia Forum. Considerations for the transition from paediatric to adult care in achondroplasia. https://achondroplasiaforum.com/wp-content/uploads/2023/06/Adult-follow-up_Melita-Irving.pdf.
38. Fredwall S, Allum Y, AlSayed M, et al. Optimising care and follow-up of adults with achondroplasia. *Orphanet J Rare Dis*. 2022;17(1):318. https://doi.org/10.1186/s13023-022-02479-3.
39. Bedeschi MF, Mora S, Antoniazzi F, et al. The clinical management of children with achondroplasia in Italy: results of clinician and parent/caregiver surveys. *J Endocrinol Invest*. 2023. https://doi.org/10.1007/s40618-023-02151-y.
40. Heath G, Farre A, Shaw K. Parenting a child with chronic illness as they transition into adulthood: A systematic review and thematic synthesis of parents' experiences. *Patient Educ Couns*. 2017;100(1):76-92. https://doi.org/10.1016/j.pec.2016.08.011.
41. Fegran L, Hall EO, Uhrenfeldt L, Aagaard H, Ludvigsen MS. Adolescents' and young adults' transition experiences when transferring from paediatric to adult care: a qualitative metasynthesis. *Int J Nurs Stud*. 2014;51(1):123-135. https://doi.org/10.1016/j.ijnurstu.2013.02.001.
42. Fredwall SO, Maanum G, Johansen H, et al. Current knowledge of medical complications in adults with achondroplasia: A scoping review. *Clin Genet*. 2020;97(1):179-197. https://doi.org/10.1111/cge.13542.
43. J P, Perpiñán S, Mayo M, et al. Estudio sobre los procedimientos profesionales, las vivencias y las necesidades de los padres cuando se les informa de que su hijo tiene una discapacidad o un trastorno del desarrollo. La primera noticia [Study on the professional procedures, experiences and needs of parents when they are told their child has a disability or developmental disorder. Breaking the news]. 2013;54:S3-9. https://doi.org/https://doi.org/10.33588/rn.54S01.2012006.
44. Fundación ALPE Achondroplasia. Health Supervision of Achondroplasia: 1 month to 1 year. https://www.fundacionalpe.org/media/Biblioteca/Manejo_clinico/Supervision-salud-1-mes-a-1-ano-1.pdf.
45. National Heart Lung and Blood Institute. Sleep Apnoea Symptoms. Updated 2023 Nov 28. Accessed Dec 6, 2023. https://www.ninds.nih.gov/health-information/disorders/sleep-apnea.
46. National Institute on Deafness and Other Communication Disorders. Ear Infections in Children. Updated 2022 Mar 16. Accessed Dec 6, 2023. https://www.nidcd.nih.gov/health/ear-infections-children.
47. Hunter AG, Bankier A, Rogers JG, Sillence D, Scott CI, Jr. Medical complications of achondroplasia: a multicentre patient review. *J Med Genet*. 1998;35(9):705-712. https://doi.org/10.1136/jmg.35.9.705.
48. Salma U, Sinha A, S S. Achondroplasia: A Rare Syndrome. CODS Journal of Dentistry. 2020;12(1):17-20. https://doi.org/10.5005/jp-journals-10063-0058.
49. Beyond Achondroplasia. Achondroplasia challenges in teenagers Accessed Dec 12, 2023. https://www.beyondachondroplasia.org/en/health/achondroplasia-challenges/teenagers.

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