Hypochondroplasia

A guide for parents

Based on the insights and experiences of families living with hypochondroplasia

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Contents

Disclaimer

This resource is provided for informational purposes only, it is not exhaustive, and is not medical advice and cannot be relied on or take the place of medical advice. Always check with your healthcare team for all healthcare, well-being, or developmental questions or concerns. Every child's needs are unique and considerations presented here may not be relevant in every instance and can vary by country and region.

Introduction

Living with hypochondroplasia

People with hypochondroplasia have a normal life expectancy and can lead full lives.[1,2] While the condition can affect growth and influence some aspects of health and development, with the right support, children with hypochondroplasia can achieve their full potential.[1,3,4]

Learning your child has a rare condition can mean becoming familiar with new terms and aspects of health and development you may not have come across before. This guide draws on insights from parents of children with hypochondroplasia to bring together information that families may find helpful, including what the condition is, how it is diagnosed, and signs and features to be aware of.

Hypochondroplasia differs from child to child,[3-5] so every family will have their own experience. Understanding the condition and what to look out for may help you feel more prepared to recognize potential health considerations early and seek support when necessary.

You may not need all of the information in this booklet right now, and some of it may never be relevant to you. But having it in one place may help you feel more informed and confident as you support your child.

Acknowledgments

This guide was developed based on the insights and experiences of families living with hypochondroplasia and patient advocacy organizations. The knowledge and expertise shared by all contributors is gratefully acknowledged.

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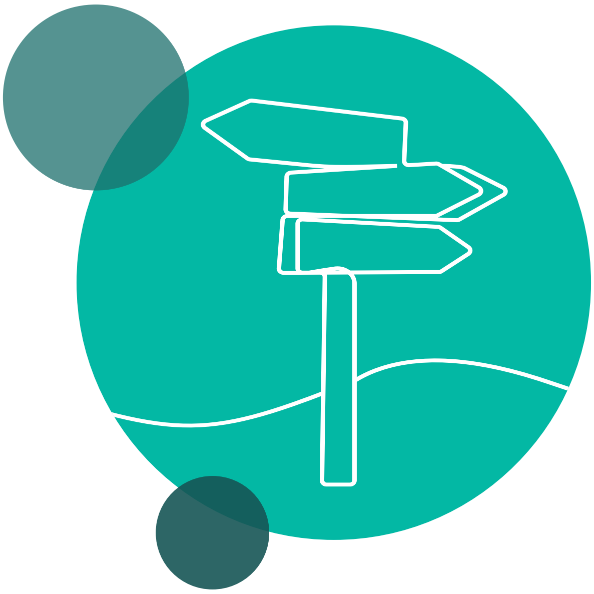
1. Understanding hypochondroplasia and where to start



What parents need to know

Hypochondroplasia is a lifelong genetic condition that affects how some bones grow.[3] It happens because of a change in a gene called FGFR3, which controls bone growth.[6] When this gene is altered, the FGFR3 protein slows the growth of bones, particularly in the arms and legs. As a result, people with hypochondroplasia are usually shorter than average and may have shorter arms and legs compared to their trunk. This is called disproportionate short stature.[3,6]

Hypochondroplasia is rare, affecting an estimated 1 in 15,000 to 40,000 people.[3,6] It can be missed or diagnosed late because the signs can be subtle and may be mistaken for typical variation in height or a different skeletal dysplasia (bone condition).[3,5,7] A genetic test can confirm the diagnosis by checking for changes in the FGFR3 gene.[3]

Most children with hypochondroplasia are born to parents of average stature. That's because the changes in the FGFR3 gene happen randomly.[3] It's therefore unlikely that average-stature parents will have more than one child with the condition.[8] If one parent has hypochondroplasia, each child has a 50% chance of having it.[3]

Hypochondroplasia affects each person differently.[3-5,9,10] The condition can be associated with some health complications, but not everyone experiences the same problems, and their severity and impact can vary from person to person.[3-5,9,10] People with hypochondroplasia have a normal life expectancy and, with the right support, can live full lives.[1,2]

Managing hypochondroplasia ideally involves a holistic, multidisciplinary approach that combines:[3,4,8,11]

- **Medical expertise** to monitor growth and potential health complications.
- **Psychosocial care** (for emotional and social well-being).
- **Educational support**.

Because hypochondroplasia is rare, many doctors may lack experience with it.[1,4] Building a trusted healthcare team and keeping thorough medical records can help ensure your child gets the care and support they need.[1]

Tips from other parents[1]

- **Try to identify specialists** who have experience with skeletal or bone growth conditions.
- **Building a small, trusted healthcare team early can be helpful**. This may include a pediatrician, a geneticist, an ear, nose, and throat (ENT) specialist, and a physiotherapist.
- **When talking to your child, simple explanations can help:** *'Your bones grow differently, so you may be shorter, but you can still do most of the things that other children do'*.
- **Keep copies of your child's medical notes and letters.** Bringing these records to appointments helps when you have consultations with new medical professionals.
- **Be prepared to advocate for your child's needs** in healthcare, at school, and in public settings. Don't let others minimize your concerns.
- **People with hypochondroplasia can lead full, meaningful, and active lives.** Help build your child's confidence and celebrate strengths.
- **Hypochondroplasia can affect more than just height.** Being aware of possible complications like learning differences or physical limitations can help ensure they are recognized early.
- **Connecting with other families** who have a child with hypochondroplasia can help you feel supported and less alone.

Questions to ask your healthcare team[1]

Do you have experience with hypochondroplasia or similar bone growth conditions?

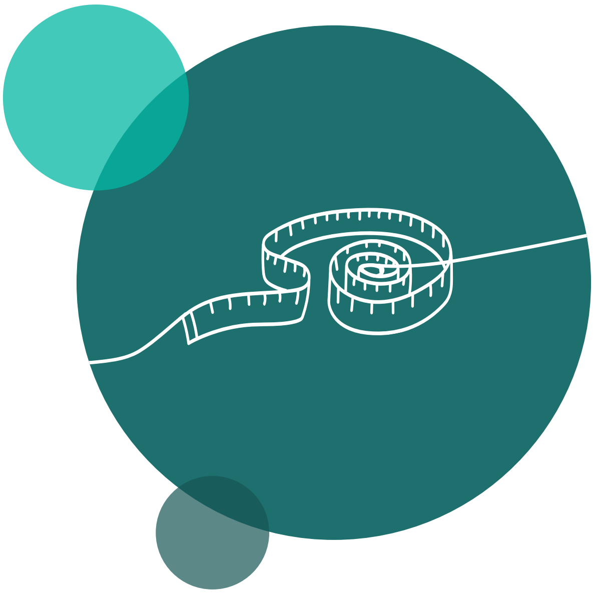
Is there a specialist clinic for skeletal dysplasia that could support our child?

Which specialists should be involved in our child's care?

Where can we find reliable information about hypochondroplasia?

Are there parent groups or community associations or resources you recommend?

2. Growth patterns and body proportions



What parents need to know

Children with hypochondroplasia usually grow differently from other children, with shorter arms and legs in proportion to their trunk.[3] Often their head may appear large compared with their body. This is called relative macrocephaly. In fact, in many cases, head circumference is within the expected range, but it seems larger because of shorter overall height.[3,12]

Growth may look typical in the first few years, but differences often become more noticeable as the child gets older.[3] In adulthood, height can range from 138 to 165 cm (4 ft 6 in to 5 ft 5 in) for men and 128 to 151 cm (4 ft 2 in to 4 ft 11 in) for women.[3]

There are specific growth charts available for children with hypochondroplasia.[6] These charts help doctors track changes in height, weight, and head circumference from infancy through to adolescence compared to other people with hypochondroplasia.[6]

Tips from other parents[1]

- **Ask your healthcare team to use hypochondroplasia-specific growth charts**, as they reflect expected growth more accurately.
- **Adapting your child's environment fosters confidence and autonomy.** Based on the experiences of other parents, small changes can make a big difference:
    - **At home:** Use light-switch extenders, step stools, lever-style taps, and lower storage for easy access.
    - **Clothing and footwear:** Look for wide-fitting shoes, as many children with hypochondroplasia have wide or thick feet. Adapt clothing with shortened sleeves or trouser legs and use easy fastenings.
    - **At school:** Work with teachers to ensure an inclusive, supportive environment that helps your child manage everyday school activities.
- **An occupational therapist, physiotherapist, or patient organization** can suggest practical adaptations to promote your child's independence and confidence.
- **Practicing how to answer questions about height** can help your child feel more confident.

Questions to ask your healthcare team[1]

Are you using the correct growth charts to track my child?

What exercises would you recommend to strengthen my child's lower limbs?

Is my child's head growth following the expected pattern?

3. Bones and joints

What parents need to know

Joint and bone issues vary widely in children with hypochondroplasia.[4] Some experience few or no problems, while others may need physiotherapy or surgical treatment to help manage their condition.[1-3] Pain can sometimes occur and may disrupt sleep after a busy day.[4]

Arms

Some children with hypochondroplasia have elbows that do not fully extend.[2,3] This may limit how effectively they can reach, for example when using the bathroom.[2]

Legs

Children with hypochondroplasia can develop bowed legs, where the bones curve outward as they grow.[3,4] Referral to an orthopedic specialist may be needed if this interferes with walking, and surgery is sometimes required if bowed legs become painful or severe.[2,3]

Back and posture

Hypochondroplasia is sometimes associated with an inward curve in the lower spine (called lumbar lordosis), which can affect posture and movement and may cause back pain.[3,13]

Spinal canal

In adulthood, some people may develop narrowing around the spinal cord (spinal stenosis). This sometimes leads to pain, numbness, or weakness in the legs.[3,14]

Joint or bone problems may require referral to an orthopedic specialist to guide appropriate management.[3] If needed, physiotherapy can help maximize mobility, and adapting your child's environment can support comfort and independence.[1,3,4]

Questions to ask your healthcare team[1]

What types of seating are recommended to support my child's head and neck in infancy?

Are there any baby carriers or equipment that may not provide adequate head and neck support?

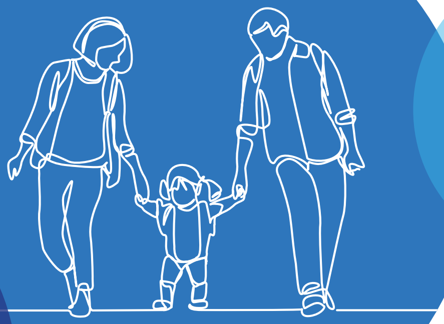
Would physiotherapy or occupational therapy be helpful for my child?

Are there any physical activities that are particularly suitable for my child?

Are there any activities that might not be suitable, for example those that involve forceful or uncontrolled neck movements?

Are there any exercises that might support my child's arm extension?

How do I manage my child's pain?



4. Ears, nose, and throat – hearing and sleeping

What parents need to know

Hearing

Infants and children with hypochondroplasia often experience recurrent ear infections and hearing difficulties. Ear problems are important to identify and manage because hearing issues can delay speech and language development, which may result in the need for additional therapy.[2,4]

Some children need small ear tubes called tympanostomy tubes or grommets placed in the eardrum to help fluid to drain from the middle ear.[2,15] This can reduce ear infections and improve hearing.[15] Tympanostomy tubes may need to be left in place until approximately 6–8 years of age.[2] Hearing aids may also be considered if required.

Sleeping

People with hypochondroplasia can develop sleep apnea, a condition in which breathing briefly stops and starts many times during the night.[3,9,16] This can affect the quality of sleep and cause daytime tiredness.[16,17] If untreated, sleep apnea can lead to problems concentrating or paying attention at school, behavioral changes such as being overactive, and affect academic performance.[16-18]

Sleep apnea can be detected through a sleep test (sleep polysomnography) and different treatment options may be considered.[3,16,17] If needed, children may wear a small mask while sleeping that is connected to a breathing support machine such as a continuous positive airway pressure (CPAP) device, which helps keep the airways open.[17,19] Tonsils or adenoids are sometimes removed surgically to improve the condition.[18]

Occasionally, the structure of the base of the skull can contribute to sleep apnea, which may require referral to a neurologist for investigation and treatment.[3,4,9]

Different specialists may be involved in the care of children with ear, nose, and throat (ENT) symptoms depending on their specific needs, including sleep specialists, ENT doctors, and neurologists.[2,3]

Tips from other parents[1]

- **Early audiology checks are important**, as ear problems are not always obvious in infants.
- **Annual hearing assessments are recommended** from 9–12 months of age.
- **Snoring, pauses in breathing, night waking, or daytime tiredness** may be signs of sleep apnea.
- **Making a short video recording of your child sleeping** can be helpful to show doctors if you are unsure about your child's breathing.
- **Sleep studies can feel stressful** but often give important information about how your child is breathing at night.

Questions to ask your healthcare team[1]

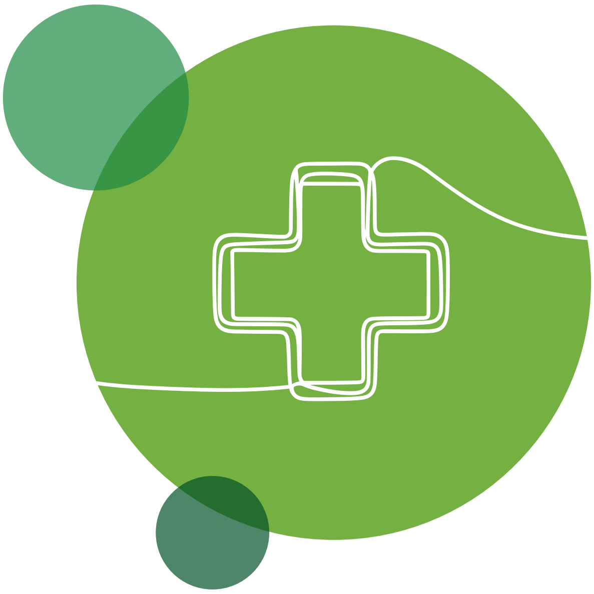
Is there fluid behind the eardrum today?

Would ear drainage tubes be helpful for my child?

Does my child need a sleep study?

What signs of sleep apnea should we watch for?

5. **Seizures and other neurological complications**



What parents need to know

Seizures

A small number of children with hypochondroplasia experience seizures.[2,4] These can occur at any age, but often happen in infancy, where the signs can be subtle, including brief pauses in breathing, staring spells, and stillness.[2,3,10,20,21]

Children with hypochondroplasia who have a seizure should have tests including an electroencephalogram (EEG) and magnetic resonance imaging (MRI), and be treated by an experienced neurologist.[2,3] Medicines for epilepsy are used to manage seizures.[2,3,21] Seizures that begin in infancy or early childhood may subside and some children may experience remission by school age.[4,10]

Hydrocephalus

A small minority of children with hypochondroplasia develop hydrocephalus that requires treatment.[2,4,9,10]

Hydrocephalus is an abnormal build-up of fluid in the brain.[22] Doctors closely monitor the circumference of the head and if it grows faster than expected or signs of fluid build-up appear (such as vomiting, irritability, poor feeding, or unusual drowsiness), referral to a neurologist may be needed and imaging scans like MRI or computed tomography (CT) undertaken to evaluate the condition.[2,3,22] If the child has symptoms of hydrocephalus, surgery to place a shunt (to drain fluid) may be considered.[2,22]

Tips from other parents[1]

- **Record any unusual episodes on your phone**, as this can help healthcare providers assess what is happening.
- **If something does not feel right**, trust your instincts and seek medical advice.
- **Families should be aware of signs that require immediate medical attention**, particularly related to fluid build-up in the brain (persistent vomiting, extreme irritability, or unusual tiredness) and seizures.

Questions to ask your healthcare team[1]

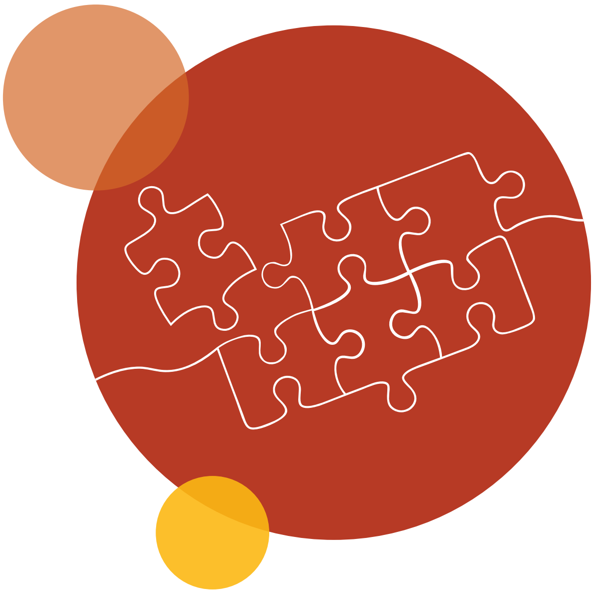
What do seizures typically look like in infants or children with hypochondroplasia?

If another episode occurs, what should we do during and after the seizure?

What symptoms should prompt us to seek urgent medical review, for example signs of hydrocephalus?

Does my child need an EEG, MRI, or other tests to check for neurological complications?

6. Developmental milestones and learning support



What parents need to know

Some children with hypochondroplasia experience developmental delays, for example reaching speech or motor milestones later than expected.[1,3,4]

Movement

For problems with gross motor skills (movements made using large muscles like those in the arms and legs), physiotherapy is recommended to maximize mobility.[3,23] Occupational therapy is advised for children experiencing difficulty with fine motor skills that affect everyday living such as feeding, grooming, dressing, and writing.[3]

Learning

Although many children with hypochondroplasia have typical intelligence, learning difficulties and developmental delays can occur, including challenges with language development and trouble concentrating.[2-4,9,10] Certain medical complications associated with hypochondroplasia can affect learning, including sleep apnea, hearing difficulties, hydrocephalus, and seizures or epilepsy.[2,4,18,20,22]

It is recommended that children with hypochondroplasia have regular developmental check-ups to help ensure that any motor, speech, and learning difficulties are identified early and supported through appropriate therapies and educational services.[3,10]

Tips from other parents[1]

- **Routine developmental assessment is important.** Request a more detailed evaluation if you notice delays, so that early intervention can support your child's progress.
- **Keep an eye on speech, gross motor development, fine motor development, and attention.** Early support can make a significant difference.
- **Hearing problems can look like learning difficulties.** Annual hearing checks are important.
- **If learning challenges arise,** ask the school about options such as individualized education programs or additional teaching support. Many families find these make a positive difference.

Questions to ask your healthcare team[1]

Are my child's developmental milestones on track for hypochondroplasia?

Which developmental assessment tools are you using (for example, the Ages and Stages Questionnaire)?

Are there any tests that would help understand what is causing my child's learning or attention difficulties?

How often should my child's hearing and vision be checked?

Are there any investigations that could be considered to exclude other causes of developmental problems such as attention-deficit/hyperactivity disorder (ADHD), attention-deficit disorder (ADD), or autism spectrum disorder (ASD)?

What interventions would my child benefit from, for example speech therapy, physiotherapy, or occupational therapy?

What information should we share with the school to help them support learning? Are there any specific educational interventions you feel my child would benefit from?

7. Emotional and mental well-being for families[1]

What parents need to know

A diagnosis of hypochondroplasia can affect the whole family in different ways. With the right support, parents and children can adapt and thrive. Looking after emotional and mental well-being is an important part of overall health.

Parents

Parenthood has ups and downs, but learning your child has a rare condition can sometimes feel overwhelming. Parents may experience a range of emotions, including shock, worry, and sadness, especially when support is hard to find. Constantly advocating across healthcare, school, and support services can be tiring. Over time, many families build strength and resilience, especially when they have clear guidance and supportive communities.

Parents with hypochondroplasia

For parents who also have hypochondroplasia, raising a child with the same condition can feel both familiar and challenging. Their personal experience helps them understand and support their child, while also bringing up memories of their own journey.

Couples

Caring for a child with hypochondroplasia can put strain on a relationship. Medical demands, stress, and financial pressures may leave less time for each other and make communication harder. Yet many couples find that facing these challenges together strengthens their bond and builds resilience.

Children

Children with hypochondroplasia may notice their differences early, which can bring sadness, worry, or frustration. Social pressures, medical appointments, and dependence on adults can add to these feelings. With supportive families and inclusive environments, children often grow in confidence and resilience.

Siblings

Brothers and sisters may feel overlooked or anxious when parents' attention is focused on medical care. They might struggle with jealousy, guilt, or pressure to be strong. With reassurance and open communication, siblings can develop empathy, resilience, and pride in their role, helping the whole family feel more connected.

Tips from other parents

Looking after yourself

- **Give yourself time to process the diagnosis – strong emotions are normal.** Focus on one step at a time to reduce feelings of being overwhelmed.
- **Only use information from trusted sources;** too much internet searching can increase your anxiety.
- **Share your feelings with someone you trust;** you do not need to carry them alone.
- **Connecting with parent groups or patient associations** (including on social media) can reduce feelings of isolation and provide practical guidance and a platform for parents to discuss their concerns and learn from others in the same situation.
- **Contact the skeletal condition patient association in your country.** These organizations are an important source of information and support and can provide valuable guidance on how to manage the challenges faced by people with short stature.
- **Find ways to nurture your relationship** and keep your partnership strong if you are co-parenting.
- **Some people will feel ashamed about not coping as well as they would like.** It's okay to ask for help from others if you are struggling.
- **If you have persistent low mood** or prolonged feelings of emotional distress, it may be helpful to talk to a psychologist or counselor.

Supporting your child with hypochondroplasia

- **Children may become aware of their differences from an early age**. Feelings of frustration, sadness, or anxiety are common and understandable.
- **Validate your child's feelings before offering reassurance.** This helps them feel heard and supported.
- **Help your child practice simple responses to insensitive questions or stares.** Practice simple phrases your child can use, such as: *'My body grows differently'*.
- **Let your child share their feelings**, even the difficult ones – listening comes first.
- **Encourage friendships** and inclusive activities where your child feels confident.
- **Work with teachers** to create a safe, supportive school environment.
- **Balance being overprotective versus fostering your child's independence.**
- **Celebrate your child's strengths and efforts** – it builds self-esteem over time.

Supporting other children in the family

- **Give siblings space** to express their own feelings without judgment.
- **Create small moments of one-to-one time** with each sibling when possible.
- **Explain the condition in simple, age-appropriate ways**, so siblings understand what is happening.
- **Look out for signs of worry, jealousy, or feeling left out**, and talk openly about them.
- **Involve siblings in positive ways:** helping, playing, or cheering each other on.

Questions to ask your healthcare team

How can we best support our child's emotional well-being?

Are anxiety or low mood common in children with hypochondroplasia?

Would counseling, play therapy, or other psychological support be helpful?

Is there a patient association or online patient community that you can recommend?

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