



# FREDERICK'S GROWTH FUND

321-829 NPO

## A Starting Guide

Learning that your baby has achondroplasia can bring many emotions at once – surprise, worry, love, protectiveness, and a thousand questions about the future. Many parents receive this diagnosis during pregnancy or shortly after birth, often without much guidance about what comes next.

The most important thing to know is that **children with achondroplasia can live full, joyful, meaningful lives**. With the right medical care, awareness, and support, most health risks can be monitored and managed effectively. You are not alone, and there is a growing community of parents, doctors, and advocates working to improve care and opportunities for children with achondroplasia.

I created this guide as a **starting point** – a practical overview of things to monitor at different stages of life, and the specialists who may be helpful along the way. Not every child will experience every challenge listed here, but knowing what to watch for can help you advocate confidently for your child's health and wellbeing.

*with love,  
Charnelle*



## Infancy & Early Childhood

(0-5 years) High-Risk Period

This early stage of life requires **the most careful monitoring**, as some children with achondroplasia can experience complications related to the brain, breathing, and spine. Early detection and specialist care can significantly reduce risks.

Area	What to Watch	Recommended Action
Brain & Breathing	Poor feeding, weak muscles, snoring, blue spells, floppy limbs.	Always cradle and support baby's neck and back. <b>NO</b> baby jumpers! Avoid impact at neck and skull base junction.  Early MRI of skull base, regular neurological check-ups, sleep study; possible surgery if severe compression.
Signs of Increased Brain Pressure	Bulging soft spot (fontanelle), enlarged pupils, vomiting, irritability, poor feeding.	Contact neurosurgeon or pediatrician immediately; urgent imaging may be needed.



# Infancy & Early Childhood (Continued)

Area	What to Watch	Recommended Action
Post-Fossa Decompression	Surgery to widen the Foramen Magnum to relieve brainstem compression. Incision infection. Fever, drowsiness, confusion, severe nausea and vomiting. Dizziness, off-balance, severe numbness or weakness in arms and legs. Enlarged pupils. Contact the Doctor immediately.	No jumping, rough play, or high-impact activity – cerebellum can protrude through widened skull opening.
Sleep Apnea	Two types: <ul style="list-style-type: none"> <li>• Central sleep apnea (brainstem compression).</li> <li>• Obstructive sleep apnea (small airways, large tonsils/adenoids, midface differences).</li> </ul>	Sleep study, ENT review, possible tonsil/adenoid removal, CPAP if needed.  Sleeping/Cot Placement: Back sleeping only (supine) not on stomach. Head neutral (no chin-to-chest, no overextension). Empty cot (no extra blankets, toys, bumpers). Avoid slumped sleep, positional asphyxia more likely due to heavier head (no car seats/bouncers for sleep).  Watch for warning signs during sleep, including snoring, noisy or labored breathing, and pauses in breathing (apnea). Other indicators may include excessive sweating, restlessness, frequent waking, or unusual sleeping positions such as arching the body or extreme head tilting.
Ears & Hearing	Frequent ear infections, delayed speech.	Regular hearing tests, treat infections promptly, grommets if needed.
Spine & Posture	No unsupported sitting. No baby jumpers. Ensure the spine is supported and straight.	Support neck/back, avoid unsupported sitting, monitor for kyphosis.
Head Growth	Rapid increase may signal hydrocephalus.	Measure head circumference weekly and plot on achondroplasia-specific chart.
Dressing Guidelines	Avoid pulling clothes over the head.  	Use bottom-up baby grows; gently put arms through sleeves first. Envelope Neckline Grow - overlapping fabric at the shoulders allows the neckline to stretch, making it easier to put on and take off by pulling it down over the baby's body rather than over the head.
Transport	Head rolling forwards compressing airway leading to positional asphyxia which is usually silent, saliva bubbles around the mouth.	Carseat to be inclined rear facing (install backseat mirror), head must be supported to avoid slumping forward/sideways, baby must be supervised, check constantly.



## Red Flags

Contact a Doctor Immediately

- Difficulty breathing or pauses in breathing
- Saliva bubbles
- Blue lips or skin
- Sudden weakness or inability to move
- Poor feeding or persistent vomiting
- Rapidly enlarging head
- Bulging fontanelle
- Enlarged pupils



## Recommended Specialists

(0-5 years)

- Genetic counselor
- Pediatrician familiar with achondroplasia
- Pediatric neurosurgeon (for foramen magnum/ hydrocephalus issues)
- Pediatric ENT (sleep apnea, tonsils, grommets)
- Pediatric orthopedist (spine, legs)
- Pediatric neurologist
- Pediatric physiotherapist



## Childhood (5–12 Years)

During this stage, the focus shifts toward **healthy development, movement, and monitoring bone and joint health.**



### Areas to monitor

- ❑ Spine and posture.
- ❑ Leg alignment.
- ❑ Growth using **achondroplasia-specific charts.**
- ❑ Maintaining a healthy weight and active lifestyle.



### Recommended Specialists

(5-12 years)

- Pediatric orthopedist (spine, legs).
- Pediatric neurologist (if neurological issues persist).
- ENT / audiologist (hearing monitoring).
- Physiotherapist (posture and strength).
- Dietitian (healthy growth and weight management).
- Genetic counselor (optional).

## Adolescence

Adolescence brings new challenges and opportunities as children grow into their independence. Emotional wellbeing, identity, and confidence become just as important as physical health.



### Areas to monitor

- ❑ Supporting self-confidence and emotional wellbeing.
- ❑ Monitoring back pain and joint health.
- ❑ Maintaining activity and healthy weight.
- ❑ Encouraging independence and self-advocacy.



### Recommended Specialists

(12-18 years)

- Orthopedist (spine and joints).
- Physiotherapist / occupational therapist.
- Psychologist or counselor (self-esteem, mental health).
- Endocrinologist if growth or puberty concerns arise.
- ENT / audiologist as needed.

Medical Journal: Health Supervision for People with Achondroplasia

Download an achondroplasia-specific growth chart



## Adulthood

Adolescence brings new challenges and opportunities as children grow into their independence. Emotional wellbeing, identity, and confidence become just as important as physical health.



### Areas to monitor

- Spine and joint health.
- Chronic back pain or arthritis.
- Maintaining healthy weight and activity levels.



### Recommended Specialists

(18+ years)

- Orthopedic surgeon (spine/joints).
- Physiotherapist / occupational therapist.
- Rheumatologist (arthritis).
- Endocrinologist (bone or hormonal health).
- Dietitian (weight management).
- Psychologist / counselor (mental health support).
- ENT / audiologist as needed.



## Key Tips for All Ages

- Avoid unsupported sitting or jumping if post-fossa decompression has been performed.
- Keep medical records organised and up to date.
- Monitor sleep, breathing, hearing, and head growth.
- Use achondroplasia-specific growth charts.
- Encourage healthy eating and regular physical activity.
- Build a supportive care team early.

## Support & Contacts

### Frederick's Growth Fund – South Africa

Frederick's Growth Fund provides guidance on medical specialists, treatment pathways, and lived-experience support for families navigating achondroplasia in South Africa.

✉ [info@fgfnpo.org](mailto:info@fgfnpo.org) • [www.fgfnpo.org](http://www.fgfnpo.org)

*healthy growth and dignified lives*