BIOMARIN®

LIVING WITH HYPOCHONDROPLASIA:

A Qualitative Exploration of Children's and Caregivers' Experiences, Challenges, and Unmet Needs

WHAT THIS STUDY WAS ABOUT

This study was about understanding what life is like for families with a child who has hypochondroplasia (HCH), a rare genetic skeletal dysplasia characterized by short stature, disproportionate limbs, and complications such as learning differences.

First, researchers talked with 9 children and young adults with HCH and 25 caregivers of children with HCH in one-on-one video interviews. After the interviews, researcher conducted focus-group discussions with 10 caregivers to learn more about topics that came up in the interviews.

This research was supported by BioMarin Pharmaceutical Inc.

We Asked:

- How do families find out their child has HCH?
- What has the care and treatment of HCH been like?
- How does HCH affect day-to-day life?

How the HCH Journey Often Begins

Common signs and symptoms include:

DURING PREGNANCY

An ultrasound reveals short long bones or halted bone growth.

AT OR SHORTLY AFTER BIRTH

The birthing team or family notices disproportionate limb length, an enlarged head, brief pauses in breathing, or seizures.

DURING INFANCY

The child shows slowed growth or develops bowed legs.

AS A TODDLER

The child misses developmental milestones such as delayed speech or walking, or caregivers sense something is not right.

When signs are recognized during pregnancy, birth, or early infancy, the diagnostic process tends to be more direct. When signs emerge later in childhood, diagnosis is more complex

What Prevents A Timely Diagnosis

- Difficulty scheduling specialist appointments or delayed test results
- Lack of achondroplasia signs and symptoms and unfamiliarity of healthcare providers with HCH
- The assumption that the child resembles smaller family members

Caregiver emotions at diagnosis:

- **Initial Surprise**
- **Uncertainty & Self-Doubt**
- **Grief & Concern**
- **Relief**
- **Ongoing Worry**

> *"You just want the best for your child, and you want your child to grow up as normal as possible … The biggest takeaway I could say is the unknowns. It's not knowing what this life would bring."*
>
> – Caregiver of a child with HCH

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Accommodations for short stature and disproportionate limb length

AT SCHOOL:

- Chair cushions for comfort
- Foot stools for support and reach
- Lowering items on walls (e.g., trays, utensils, passes)
- Notes written lower on whiteboards for visibility

HOME:

- Stools to help reach high cabinets
- "Grab-It" tools to pick up or reach things
- Homes designed with all bedrooms and living spaces on the first floor

VEHICLES:

- Booster seats
- Adjusted seatbelt mechanisms for safety
- Pedal extenders to make it easier to drive

CLOTHING:

- Supra-Malleolar Orthoses (SMOs) to support the feet and ankles
- Sandals worn year-round to fit wide feet comfortably
- Altering clothing to fit better, such as shortening pants or sleeves

Emotional Impacts

Many participants described the emotional impact of short stature, especially insecurity about their height. Several had experienced bullying at school.

Caregivers expressed concerns, especially for their boys with HCH, of societal pressures to be tall.

Caregivers described the importance of instilling a sense of confidence in their children.

While not everyone will face every complication, the cumulative impact of multiple HCH challenges can be overwhelming

Learning Differences and Challenges:

- Developmental Delays
- Mental Health Concerns
- Sleep Apnea

Epilepsy or Seizures:

- Joint Pain
- Ear Infections and Hearing Loss
- Weight Management

> *"She had one seizure as an infant but no other seizures... She definitely struggles with anxiety, a lot of anxiety and sometimes depression. And she recently said she feels sad a lot, and she shoves it down. And so, we're working on that. She has mild sleep apnea. We're probably due for another sleep study. She does have quite a bit of joint pain. And what I was saying earlier, we can't go on really long walks because she almost always has severe pain afterwards. So, she has a lot of joint pain. She had a ton of ear infections until we got tubes."*
>
> – Caregiver of a child with HCH

Needed Health System Improvements:

- Increase provider education so more healthcare professionals recognize the signs of HCH and understand how to manage it
- Develop clear care pathways and easy-to-understand materials to help families navigate next steps after diagnosis
- Improve care coordination across specialists to reduce gaps, confusion, and delays in treatment
- Make genetic testing easier to access

Advice caregivers have for those newly diagnosed:

- Be prepared to advocate for your child's needs in healthcare, at school, and in public settings.
- Don't let others minimize your concerns.
- Explore genetic testing options—these are available and can support diagnosis and care planning.
- People with HCH lead full, meaningful, and active lives. Encourage confidence and celebrate strengths.
- Sometimes HCH affects more than just height. Be aware of possible complications like learning differences or physical limitations.
- Your child may look younger than peers, especially as height differences become more pronounced with age