HYPOCHONDROPLASIA AND EAR, NOSE & THROAT ISSUES

Academic papers, medical conference abstracts and clinical guidance

Evidence review prepared for the Hypochondroplasia Foundation

Search updated: 17 July 2026

Scope: hypochondroplasia-specific and mixed-cohort evidence relating to otitis media, middle-ear dysfunction, hearing, ventilation tubes, adenoids, tonsils, upper-airway obstruction, sleep-disordered breathing, palate and related craniofacial findings.

This report is an evidence map, not a clinical guideline or individual medical advice.

Executive summary

The HCH-specific ENT evidence base is small but no longer negligible. The most consistently identified issues are recurrent otitis media, persistent middle-ear dysfunction, conductive or mild hearing difficulty, tympanostomy procedures, obstructive sleep apnoea, and adenotonsillar disease or surgery. Recent datasets provide the first reasonably sized HCH-specific estimates, but the literature remains fragmented and methodologically heterogeneous.

- The strongest current HCH-specific dataset is the 2026 ACCEL interim cohort of 64 molecularly confirmed children.
- ACCEL recorded otitis media in 21.9%, obstructive sleep apnoea in 12.5%, adenoidal hypertrophy in 6.3%, ear-tube insertion in 15.6%, adenoidectomy in 21.9%, tonsillectomy in 9.4%, and myringotomy in 6.3%.
- A 2025 English matched-cohort abstract found significantly higher overall ENT event rates in children coded as having HCH than in controls, with rate ratios of 1.84 at ages 0-10 and 5.21 at ages 11-18.
- A Danish chart review reported conductive hearing loss in 3 of 20 people with HCH, although its recurrent-otitis table contains an internal numerical inconsistency.
- Qualitative HCH research documents recurrent ear infections, multiple sets of tubes, mild hearing difficulties, speech-language effects, snoring, breathing pauses and sleep-apnoea treatment from the family perspective.
- Many reviews repeat ENT concerns without adding new HCH patient data; achondroplasia findings must not be treated as HCH prevalence estimates.

Bottom-line interpretation

Scientifically defensible wording: Children with hypochondroplasia may have an increased burden of recurrent otitis media, middle-ear procedures, conductive or mild hearing difficulty, obstructive sleep apnoea and adenotonsillar interventions. These issues are variable and are not present in every child. Their true prevalence, mechanisms, age course and optimal surveillance strategy have not yet been established through a dedicated prospective HCH otolaryngology study.

Scope and search approach

The review sought publications and conference materials containing people with hypochondroplasia and an identifiable ear, nose, throat, hearing or upper-airway finding. Searches combined hypochondroplasia or FGFR3 with terms including otitis, hearing loss, tympanometry, middle ear, ventilation tubes, myringotomy, adenoid, tonsil, nasal obstruction, sleep apnoea, upper airway, cleft palate, craniosynostosis, ENT and otolaryngology.

- Included: peer-reviewed human studies with direct HCH data; mixed skeletal-dysplasia studies that included HCH; medical conference abstracts or posters; HCH clinical reviews and management resources; unusual craniofacial case reports with potential ENT relevance.
- Separated: original patient data versus statements repeated in introductions or reviews.
- Not counted as HCH prevalence evidence: achondroplasia-only ENT studies, animal studies, general FGFR3 biology papers, marketing materials and non-medical webpages.
- Limitations: the search was not registered as a formal systematic review; obscure non-indexed national abstracts may have been missed; several conference outputs report the same underlying cohort.

Evidence classification used in this report

Category	Meaning	How to use it
Direct HCH data	HCH results are reported separately or the cohort is entirely HCH.	Suitable for describing HCH findings; still interpret design limitations.
Mixed cohort	HCH participants are included but results are not separated.	Shows inclusion and plausibility, not HCH prevalence.
Recognition / guidance	A review or guidance document identifies the issue without new patient data.	Useful for clinical context; do not double-count as independent evidence.

Key quantitative HCH findings

Finding	Estimate	Source / cohort	Interpretation
Otitis media	14/64 (21.9%)	ACCEL 2026; molecularly confirmed children	Cumulative recorded medical history at interim analysis
Conductive hearing loss	3/20 (15%)	Danish chart review, 2017	Small retrospective cohort
Obstructive sleep apnoea syndrome	8/64 (12.5%)	ACCEL 2026	Recorded diagnosis; no PSG detail in abstract
Adenoidal hypertrophy	4/64 (6.3%)	ACCEL 2026	Recorded diagnosis
Ear-tube insertion	10/64 (15.6%)	ACCEL 2026	Procedure history
Myringotomy	4/64 (6.3%)	ACCEL 2026	Procedure history
Adenoidectomy	14/64 (21.9%)	ACCEL 2026	Procedure history; exceeds recorded adenoidal-hypertrophy diagnosis
Tonsillectomy	6/64 (9.4%)	ACCEL 2026	Procedure history
Overall ENT events vs controls, age 0-10	RR 1.84 (95% CI 1.34-2.52)	English CPRD matched cohort, 2025	Aggregated ENT category, not diagnosis-specific
Overall ENT events vs controls, age 11-18	RR 5.21 (95% CI 3.24-8.38)	English CPRD matched cohort, 2025	Aggregated ENT category, not diagnosis-specific

Do not pool these estimates as a meta-analysis. They measure different outcomes: diagnoses, procedures, administrative event rates, screening findings and caregiver experiences.

1. Original peer-reviewed human studies

1.1. Schlüter et al. - sleep-related respiratory disturbances in FGFR3 skeletal dysplasia | *Georgian Medical News, 2011 | Mixed cohort*

Population: 24 children: 22 with achondroplasia and 2 with hypochondroplasia.

ENT / sleep findings: In the combined cohort, 10/24 had abnormal polysomnography, 8/24 obstructive sleep apnoea syndrome, 11/24 hypertrophied tonsils, and 8/24 disturbed nasal breathing. ENT procedures including tonsillar and adenoidal surgery were described.

HCH interpretation: The two HCH participants were not reported separately. The percentages must not be quoted as HCH prevalence.

Evidence value: Confirms that HCH was represented in an FGFR3-specific paediatric sleep and upper-airway cohort, but contributes no separable HCH estimate.

1.2. Tunkel et al. - hearing screening in children with skeletal dysplasia | *Archives of Otolaryngology-Head & Neck Surgery, 2011 | Mixed cohort*

Population: 58 children with skeletal dysplasia, including one six-year-old girl with HCH; 56 completed hearing screening.

Overall findings: More than one quarter failed hearing screening and middle-ear dysfunction was common across the cohort. The single HCH participant's result was not reported separately.

Evidence value: Useful for showing how little HCH-specific audiology data existed; not an HCH prevalence study.

Conference history: Presented at the American Society of Pediatric Otolaryngology annual meeting in Chicago in April 2011.

1.3. Tunkel et al. - hearing loss in skeletal dysplasia patients | *American Journal of Medical Genetics Part A, 2012 | Small HCH subgroup*

Population: 110 people with skeletal dysplasias completed screening; the non-achondroplasia subgroup included HCH.

HCH-specific result: The paper explicitly states that hearing was normal in those with hypochondroplasia.

Interpretation: Important negative evidence: hearing loss is not inevitable in HCH. The exact number of HCH participants is not clear from the abstract and the subgroup was small. A single screening visit cannot exclude intermittent conductive loss or recurrent otitis.

Overlap: This study likely overlaps substantially with the 2011 Little People of America hearing-screening cohort and should not be treated as wholly independent evidence.

1.4. Doherty et al. - Danish achondroplasia and hypochondroplasia cohort | *Journal of Rare Diseases Research & Treatment, 2017 | Direct HCH subgroup*

Population: 48 people with achondroplasia and 20 mutation-verified people with HCH; retrospective chart review across three Danish clinics.

HCH-specific result: Conductive hearing loss was reported in 3 of 20 people with HCH (15%). Recurrent or multiple otitis media and sleep apnoea were also identified in the HCH group.

Data-quality caution: The online table appears to state that 15 HCH participants had multiple otitis media while labelling this as 40%. With a denominator of 20, 15 would equal 75%. The presence of recurrent otitis can be cited, but the exact percentage should not be used without clarification from the authors or source dataset.

Evidence value: One of the clearest older HCH-specific sources for conductive hearing loss, but small and retrospective.

1.5. Oehrlein et al. - living with hypochondroplasia | *Molecular Genetics & Genomic Medicine, 2025 | Direct qualitative HCH data*

Population: Nine children or young adults with physician-confirmed HCH, 25 caregivers, and two caregiver focus groups involving ten caregivers.

Ear and hearing findings: Families described recurrent ear infections, mild hearing difficulties, some children needing multiple sets of ventilation tubes, and hearing-related effects on speech or language.

Sleep and airway findings: Families described snoring, observed breathing pauses, mild to severe sleep apnoea, stressful sleep-study experiences, and surgical treatment that improved breathing or sleep in some children.

Interpretation: Rich direct evidence of lived experience and clinical burden. It is qualitative and should not be converted into prevalence percentages.

1.6. Dauber et al. - vosoritide treatment for children with HCH | *eClinicalMedicine, 2024 | Recognition, not ENT outcomes*

Relevance: The paper identifies sleep apnoea and recurrent otitis media as recognised HCH complications in its background discussion.

Limitation: The trial focused on growth, safety and skeletal outcomes and did not provide a dedicated analysis of HCH otitis, hearing, adenotonsillar disease or sleep-apnoea mechanisms.

How to cite: Use as evidence that contemporary HCH research recognises these complications, not as a prevalence source.

2. Medical conference abstracts and posters

2.1. Cheung et al. - medical impact of HCH in English children | *ECE-ESPE Joint Congress, 2025 | Direct coded HCH cohort*

Population: 225 children coded as having HCH and 1,067 matched controls in linked English electronic medical records, 1998-2019.

ENT result: Overall ENT event rates were higher in the HCH cohort: RR 1.84 (95% CI 1.34-2.52) at ages 0-10 and RR 5.21 (95% CI 3.24-8.38) at ages 11-18.

Strength: Largest identified comparative paediatric dataset reporting an ENT body-system category.

Limitations: The abstract does not break ENT events into diagnoses. Case identification included a specific HCH code or broader Q77.4 coding after exclusions, so some misclassification is possible. Event rates are not the same as the proportion of children affected.

2.2. Dauber et al. - ACCEL interim observational study | *ACMG 2026, Genetics in Medicine Open P111 | Direct molecularly confirmed HCH cohort*

Population: 64 children with molecularly confirmed HCH, aged approximately 2.5-13.2 years, from 25 sites in Asia, Australia, Europe and North America; 55/64 had p.Asn540Lys.

Diagnoses: Otitis media 14/64 (21.9%); obstructive sleep apnoea syndrome 8/64 (12.5%); adenoidal hypertrophy 4/64 (6.3%).

Procedures: Adenoidectomy 14/64 (21.9%); ear-tube insertion 10/64 (15.6%); tonsillectomy 6/64 (9.4%); myringotomy 4/64 (6.3%).

Evidence value: Currently the clearest multinational, molecularly confirmed HCH dataset spanning ENT diagnoses and procedures.

Limitations: Interim observational data based on medical history. It does not provide audiometry, tympanometry, polysomnography details, annual incidence or variant-specific ENT outcomes. The study was sponsored by BridgeBio Pharma.

2.3. ACCEL at ICCBH 2026 | *International Conference on Children's Bone Health, Poster P170 | Duplicate dataset*

Content: Reports the same interim ACCEL cohort of 64 children and the same ENT diagnoses and procedures as the ACMG P111 publication.

Interpretation: A separate conference presentation but not an independent cohort; do not double-count.

2.4. Tunkel et al. at the American Society of Pediatric Otolaryngology | *ASPO Annual Meeting, 2011 | Earlier presentation*

Content: Conference presentation of the hearing-screening work later published in Archives of Otolaryngology-Head & Neck Surgery.

Interpretation: Included one child with HCH, but did not provide a separate HCH result; not an additional dataset.

3. Rare craniofacial and palate case reports

These reports concern anatomy relevant to ENT and craniofacial care, but they do not establish routine HCH complications.

3.1. Angle et al. - molecularly proven HCH with cloverleaf skull deformity | *Clinical Genetics, 1998 | Exceptional case*

Finding: A cloverleaf skull / severe craniosynostosis phenotype was reported in a person with molecularly proven HCH.

Interpretation: Historically important unusual association, not evidence of a common ENT or airway phenotype.

3.2. González-del Angel et al. - craniosynostosis and cleft palate in a Mexican family | *American Journal of Medical Genetics Part A, 2018 | Family case report*

Finding: HCH co-occurred with craniosynostosis and cleft palate in siblings from one family.

ENT relevance: Cleft palate can affect Eustachian-tube function, otitis media, conductive hearing, speech resonance and swallowing.

Interpretation: The authors presented the association as unique. One family cannot establish that cleft palate is a characteristic HCH manifestation.

3.3. Jumayeva et al. - ERF-related craniosynostosis in a patient with HCH | *Cleft Palate-Craniofacial Journal, online 2025 / print 2026 | Dual diagnosis*

Finding: A patient with HCH and multiple-suture craniosynostosis was found to have ERF-related craniosynostosis in addition to HCH.

Interpretation: Evidence for a dual molecular diagnosis, not for craniosynostosis as a usual feature of HCH.

4. Reviews and clinical guidance that identify ENT concerns

4.1. Bober et al. - Hypochondroplasia, GeneReviews | *Updated 2025*

Relevance: Identifies obstructive apnoea as possible in HCH, recommends assessment for sleep-apnoea symptoms, developmental and speech-language surveillance, and audiological evaluation when hearing or speech concerns arise. Authoritative review; no new prevalence dataset.

4.2. Pauli - Hypochondroplasia: Natural History and Management | *Clinical monograph*

Relevance: States that many infants and young children with HCH may develop recurrent or persistent middle-ear dysfunction with conductive hearing loss, at lower risk than achondroplasia, and recommends behavioural audiometry and tympanometry. Influential specialist guidance but not a conventional peer-reviewed cohort.

4.3. Lyford-Pike, Hoover-Fong and Tunkel - Otolaryngologic Manifestations of Skeletal Dysplasias in Children | *Otolaryngologic Clinics of North America, 2012*

Relevance: Broad ENT review including skeletal dysplasias and mentioning HCH. Most detailed evidence concerns achondroplasia and other conditions; do not infer HCH prevalence.

4.4. Savarirayan et al. - craniofacial best-practice guidelines in skeletal dysplasia | *Orphanet Journal of Rare Diseases, 2021*

Relevance: Covers hearing surveillance, middle-ear disease, sleep-disordered breathing, upper-airway obstruction, adenotonsillar assessment and anaesthetic considerations across skeletal dysplasias. HCH is within scope but the evidence is not HCH-specific.

4.5. Kim and Ko - clinical management of FGFR3-related skeletal dysplasia | *Annals of Pediatric Endocrinology & Metabolism, 2022*

Relevance: Includes HCH within the FGFR3 spectrum and discusses respiratory, sleep and upper-airway complications. Most detailed ENT guidance derives from achondroplasia.

4.6. Irving et al. - pathways to facilitate early recognition and diagnosis of HCH | *Advances in Therapy, 2026*

Relevance: Identifies otitis media and speech-language delay among clinical features that may contribute to HCH recognition. Diagnostic pathway, not an ENT prevalence study.

5. Publications that mention ENT issues without new ENT data

Several HCH papers repeat that otitis media, conductive hearing loss or sleep apnoea may occur, often citing GeneReviews, the Danish cohort or broader skeletal-dysplasia literature. Repetition should not be mistaken for multiple independent datasets.

5.1. Xie et al. - failure to diagnose HCH by prenatal diagnosis | *BMC Pediatrics, 2023*

Assessment: The introduction lists sleep apnoea, upper-airway stenosis and otitis among possible FGFR3-related complications; the case itself does not provide substantive HCH ENT data.

5.2. Demuynck et al. - infigratinib in an HCH mouse model | *Journal of Bone and Mineral Research, 2025*

Assessment: The human-disease background mentions otitis and speech-language issues; the work is preclinical and contains no human ENT outcomes.

5.3. Other genetics, growth and therapeutic papers | *Various*

Assessment: Background sections may mention otitis media or sleep apnoea. Unless patient-level ENT findings are reported, they should be treated as recognition only.

6. Synthesis by clinical domain

6.1. Recurrent otitis media and middle-ear dysfunction

- Direct evidence includes ACCEL otitis media at 21.9%, ear-tube insertion at 15.6% and myringotomy at 6.3%.
- The Danish cohort and qualitative family research independently identify recurrent ear infections and persistent middle-ear burden.
- The literature does not yet describe age at first episode, infections per year, effusion duration, recurrence after tube extrusion, cholesteatoma risk or variant-specific risk.

6.2. Hearing loss

- Conductive hearing loss occurred in 3/20 people with HCH in the Danish cohort.
- Qualitative research identifies mild hearing difficulties and possible speech-language consequences.
- A small screening subgroup had normal hearing, supporting substantial variability and showing that hearing loss is not universal.
- There is little evidence for a characteristic sensorineural hearing-loss phenotype in typical HCH.

6.3. Sleep-disordered breathing and upper-airway disease

- ACCEL recorded obstructive sleep apnoea in 12.5%, adenoidal hypertrophy in 6.3%, adenoidectomy in 21.9% and tonsillectomy in 9.4%.
- Families describe snoring, breathing pauses, sleep studies and improvement after surgery in some cases.
- HCH publications rarely separate adenotonsillar obstruction from obesity, craniofacial airway dimensions, reduced tone, central apnoea or cervicomedullary mechanisms.

6.4. Nose, sinus, throat, larynx and swallowing

- Evidence for HCH-specific chronic rhinosinusitis, allergic rhinitis, septal abnormality, turbinate hypertrophy or recurrent epistaxis is insufficient.
- Apart from adenotonsillar disease and unusual cleft-palate reports, no convincing HCH series were identified for laryngomalacia, tracheomalacia, vocal-cord dysfunction, subglottic stenosis, chronic dysphagia or aspiration.
- Findings from severe FGFR3 disorders or achondroplasia should not automatically be attributed to HCH.

7. Research and funding priorities

The literature supports a strong case for a dedicated multicentre HCH ENT and sleep study. A practical protocol could combine:

- Otoscopy and structured history of acute otitis, otitis with effusion and tube procedures
- Tympanometry and age-appropriate behavioural audiometry, with bone-conduction testing where indicated

- Speech-language screening and school-impact measures
- Sleep questionnaires, nocturnal oximetry and polysomnography where clinically indicated
- ENT examination of nasal obstruction, adenoids and tonsils
- Documentation of obesity, craniofacial phenotype and neurological or foramen-magnum findings
- FGFR3 variant, age, sex, geography and access-to-care variables
- Longitudinal follow-up to distinguish intermittent childhood disease from persistent impairment

Potential grant-ready research questions

- What proportion of children with molecularly confirmed HCH have persistent middle-ear effusion or measurable conductive hearing loss?
- At what ages are otitis, tube insertion, adenoidectomy, tonsillectomy and obstructive sleep apnoea most common?
- Do ENT and sleep outcomes differ by FGFR3 variant, including p.Asn540Lys versus other variants?
- Does early audiology surveillance reduce speech-language or educational impact?
- Which children with HCH should receive routine polysomnography rather than symptom-triggered assessment?
- How do HCH-specific ENT outcomes compare with matched population controls and with achondroplasia?

8. Recommended wording for Foundation materials

Evidence-based wording

Some children with hypochondroplasia experience recurrent ear infections, persistent middle-ear fluid, mild or conductive hearing difficulties, sleep-disordered breathing or obstructive sleep apnoea. Some require ventilation tubes, adenoidectomy or tonsillectomy. These issues vary widely and do not occur in everyone. Because dedicated HCH studies remain limited, hearing, speech-language development and sleep symptoms should be discussed with the child's clinical team and assessed according to individual needs.

Avoid: claiming that all children with HCH need tubes, routine surgery, routine MRI or routine polysomnography; quoting achondroplasia prevalence as though it were HCH data; describing hearing loss or sleep apnoea as inevitable.

9. Bibliography

Core HCH and mixed-cohort sources are listed first, followed by guidance and related craniofacial publications. Links are included for verification and access where available.

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Appendix: interpretation safeguards

- Achondroplasia-only studies are highly relevant to hypothesis generation and clinical awareness, but they are not HCH prevalence studies.
- The ACCEL P111 publication and ICCBH P170 poster report the same interim cohort and should be counted once.
- The two Tunkel publications probably overlap substantially and should not be assumed to be independent samples.
- The English CPRD abstract measures event rates within an aggregated ENT category, not individual diagnoses or unique patients affected.
- Procedure frequency may reflect access to specialist care, local thresholds for surgery, age distribution and retrospective documentation, not only biological disease burden.
- An absence of HCH-specific evidence for a symptom does not prove that the symptom never occurs; it means the current literature cannot establish a characteristic association.

Prepared as a literature evidence map. Clinical decisions should be made by appropriately qualified medical professionals using individual assessment and current local guidance.