ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25jun1833

Radiologic Differentiation of Hypochondroplasia from Similar Skeletal Dysplasias: A Pictorial Review

Dr. Mrudula R[1]*; Dr. B.K.D. Prasad[2]; Dr. Akanksh Chokkapu[3]

[1,2,3] MD, Department of Radiology, NRI Institute of Medical Sciences
Visakhapatnam, Andhra Pradesh, India

Corresponding Author: Dr. Mrudula R, MD*

Publication Date: 2025/07/05

Abstract: Hypochondroplasia (HCH), a mild skeletal dysplasia, presents with disproportionate short stature and subtle radiological features, often overlapping with achondroplasia and other dysplasias. Radiographic skeletal surveys are the primary diagnostic tool, revealing shortened long bones, narrowed lumbar interpedicular distances, and squared ilia. Prenatal ultrasound aids early detection, though definitive diagnosis is challenging due to phenotypic variability and the absence of a pathognomonic sign. This pictorial review synthesizes HCH's imaging features in a single patient, emphasizing differentiation through illustrative figures and diagnostic pearls, guiding radiologists in accurate diagnosis and management.

Keywords: *Hypochondroplasia, Skeletal Dysplasia, Radiology, Skeletal Survey, Achondroplasia, Differential Diagnosis, Adult Radiology.*

How to Cite: Dr. Mrudula R; Dr. B.K.D. Prasad; Dr. Akanksh Chokkapu (2025) Radiologic Differentiation of Hypochondroplasia from Similar Skeletal Dysplasias: A Pictorial Review. *International Journal of Innovative Science and Research Technology*, 10(6), 2582-2587. https://doi.org/10.38124/ijisrt/25jun1833

I. INTRODUCTION

Hypochondroplasia (HCH) is an autosomal dominant skeletal dysplasia characterized by disproportionate short stature, shortened limbs, and a stocky build [1]. HCH is milder than achondroplasia (ACH), with subtle clinical and radiological features that complicate diagnosis [2]. This review illustrates findings in a 44-year-old male with HCH, diagnosed based on clinical and radiological features. Diagnosis may be delayed until adulthood due to subtle presentation, with adult heights ranging from 138–165 cm (men) and 128–151 cm (women) [1]. Life expectancy is normal.

The lack of a single pathognomonic sign and phenotypic overlap with ACH necessitate a comprehensive radiological approach [3]. This pictorial review outlines HCH's imaging features, diagnostic modalities, and differentiation strategies, integrating illustrative figures from a single patient to guide radiologists and clinicians.

II. CLINICAL BACKGROUND

HCH presents with mild short stature, limb-to-trunk disproportion, macrocephaly, broad hands/feet, mild brachydactyly, and lumbar lordosis [1,2]. Less common findings include scoliosis, genu varum, and, in adults, osteoarthritis and spinal stenosis due to lifelong skeletal changes [1]. Phenotypic variability complicates diagnosis, requiring integration of clinical and radiological data [3,7]. In our patient, a 44-year-old male, these features were evident, including disproportionate short stature, rhizomelic limb shortening, and lumbar lordosis, leading to a clinical and radiological diagnosis of HCH.

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25jun1833

Fig 1: Clinical Photograph of a 44-Year-Old Male with HCH.

III. IMAGING FEATURES BY MODALITY

➢ *Skeletal Survey*

A radiographic skeletal survey is the primary diagnostic tool for HCH, including anteroposterior (AP) and lateral views of the spine, AP pelvis, limbs, hands, and a lateral skull [1,8]. In our patient, these radiographs revealed characteristic HCH abnormalities, with additional degenerative changes consistent with his age (44 years) [2].

➢ *Prenatal Imaging*

Prenatal ultrasound may detect mild limb shortening and increased head circumference, but definitive diagnosis before 26 weeks' gestation is challenging [12]. While not applicable to our patient's adult diagnosis, this modality is relevant for early detection in suspected cases.

IV. REGIONAL RADIOLOGIC FINDINGS

➢ *Skull*

HCH features macrocephaly with a broad, high skull but normal craniofacial morphology, lacking ACH's midface retrusion and frontal bossing [1,9]. In our patient, skull radiographs at age 44 confirmed macrocephaly.

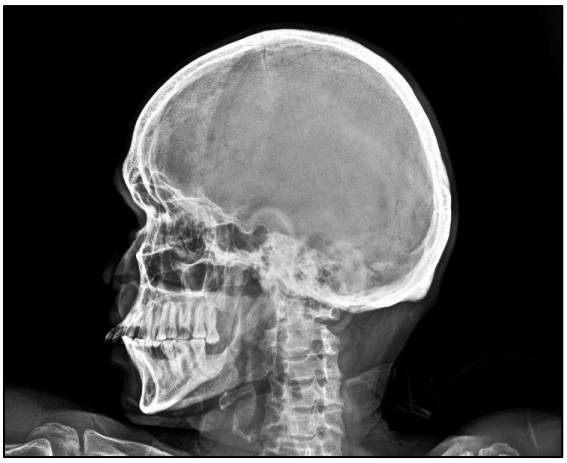

Fig 2: Lateral skull radiograph of a 44-year-old male with HCH.

➢ *Spine*

Spinal radiographs in our patient showed narrowed lumbar interpedicular distances, unlike normal caudal widening [1,8]. Anterior-posterior pedicle with mild degenerative changes (e.g., osteophytes) due to age-related wear [11]. These findings, visualized on AP and lateral views, confirmed HCH [8].

Mild scoliosis and lumbar stenosis were present, consistent with adult HCH complications, but did not require surgical intervention [13].

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25jun1833

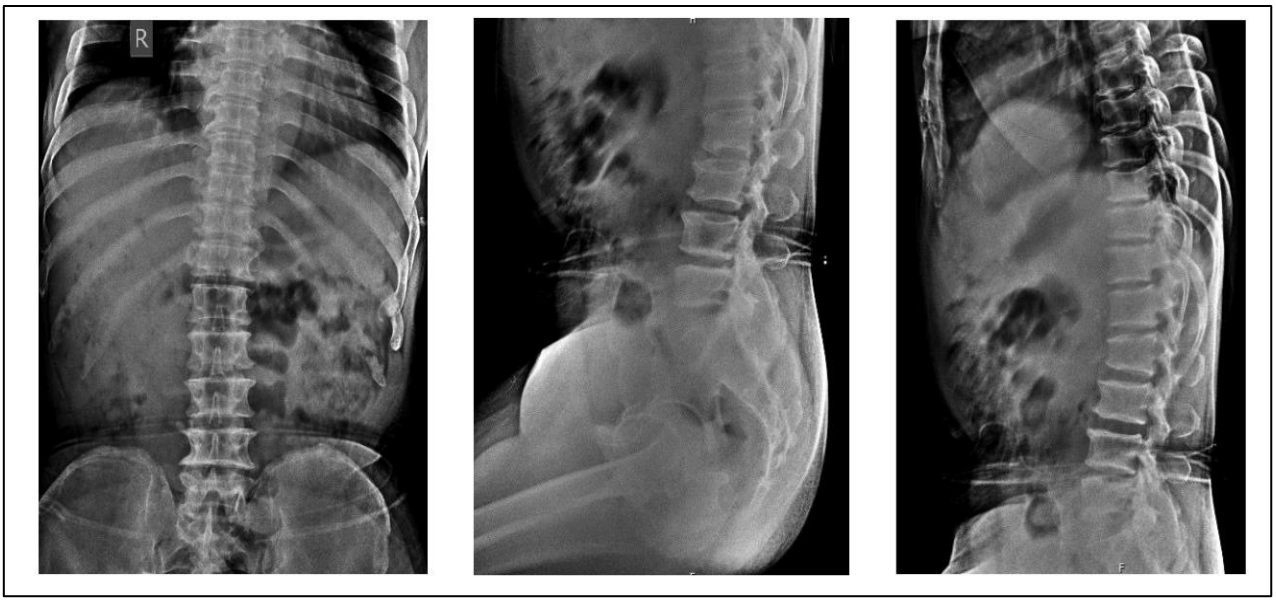

Fig 3**:** AP and Lateral Lumbar Spine Radiographs in a 44-Year-Old Male with HCH.

➢ *Pelvis*

Pelvic radiographs reveal squared, shortened ilia, low sacral articulation, horizontal orientation, and flattened acetabular roofs [1,8]. In our patient, these findings were prominent at age 44, with mild osteoarthritic changes in the hip joints, milder than ACH's "champagne glass" pelvis [14].

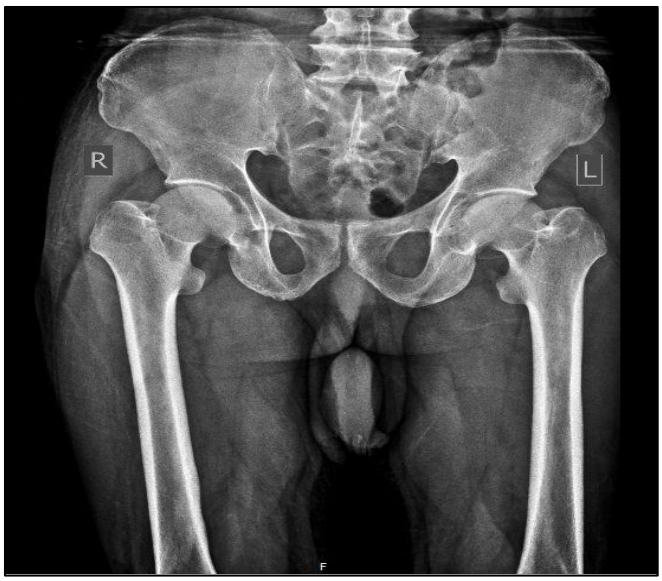

Fig 4: AP Pelvic Radiograph in a 44-Year-Old Male with HCH.

➢ *Limbs*

Long bones exhibit generalized shortening with mild metaphyseal flare, particularly in femora and tibiae [1,8]. Our patient showed rhizomelic disproportion, a short, broad femoral neck, and relative fibular elongation at age 44, with early osteoarthritic changes in the knees [2]. Hands and feet showed mild brachydactyly, with limited elbow extension [1].

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25jun1833

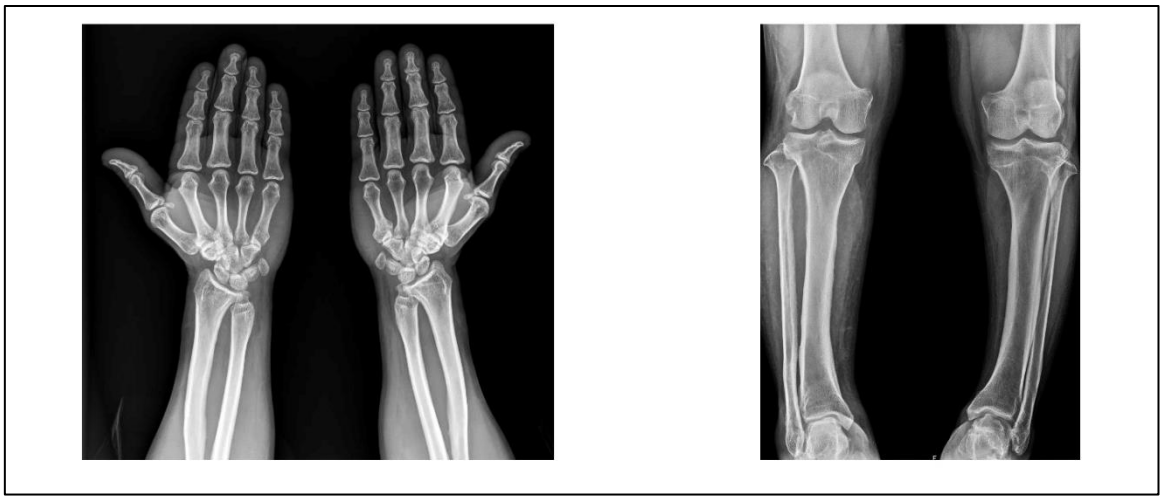

Fig 5: AP Radiograph of Upper and Lower Limbs in a 44-Year-Old Male with HCH.

V. IMAGING PEARLS AND PITFALLS

- **Pearl**: A comprehensive skeletal survey is essential, as isolated findings are nonspecific [8]. Integration of spine, pelvis, and limb findings ensures accurate diagnosis [1].
- **Pitfall**: Subtle features in childhood may delay diagnosis until adulthood, as in our patient [2]. Serial imaging may be needed to confirm HCH [8].
- **Pitfall**: Overlap with ACH requires evaluation of lumbar interpedicular distances and craniofacial features [14].

What the Radiologist Needs to Know

- **HCH Features**: Shortened femora, narrowed interpedicular distances in this patient [1].
- **ACH Differentiation**: Normal craniofacial features, milder stenosis [14].
- **Adult Presentation**: Osteoarthritis and stenosis may complicate diagnosis at age 44 [2,8].

VI. DIFFERENTIAL DIAGNOSIS

HCH must be distinguished from ACH and other skeletal dysplasias. Table 1 summarizes key radiological discriminators, with our patient's findings highlighting HCH's milder phenotype.

Table 1: Radiological Differentiation of Hypochondroplasia and Other Skeletal Dysplasias

Feature	Hypochondroplasia (HCH)	Achondroplasia (ACH)	Pseudoachondroplasia	Leri-Weill Dyschondrosteosis
Spine	Narrowed/unchanged interpedicular distance [1]	Progressive caudal narrowing, bullet-shaped vertebrae [13]	Irregular epiphyses, platyspondyly [14]	Normal or mild changes [14]
Pelvis	Squared ilia, flattened acetabular roof [8]	"Champagne glass" pelvis, rounded ilia [14]	Severe hip dysplasia [14]	Normal or mild changes [14]
Skull	Macrocephaly, normal craniofacial features [1]	Frontal bossing, midface hypoplasia [9]	Normal skull [14]	Normal skull [14]
Limbs	Shortened bones, mild metaphyseal flare [8]	Severe rhizomelic shortening, trident hand [14]	Irregular epiphyses, flared metaphyses [14]	Mesomelic shortening, Madelung deformity [14]
Complications	Mild spinal stenosis, osteoarthritis [1]	Severe spinal stenosis, hydrocephalus [13]	Severe osteoarthritis [14]	Wrist pain, deformity [14]

Other differentials include mild mesomelic dwarfism and mucopolysaccharidoses [14]. A comprehensive skeletal survey is critical to identify HCH's specific pattern [8].

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25jun1833

VII. CONCLUSION AND TEACHING POINTS

HCH's radiological diagnosis relies on a comprehensive skeletal survey. In our patient, a 44-year-old male, this modality confirmed HCH and identified mild osteoarthritis and stenosis. Its milder phenotype and overlap with ACH necessitate careful differentiation.

➢ *Summary*

- HCH in this patient showed subtle radiological features, including macrocephaly and shortened limbs.
- Comprehensive skeletal survey confirmed diagnosis at age 44.
- Radiographs identified mild osteoarthritis and stenosis, guiding management .

➢ *Teaching Points*

- HCH presents with shortened long bones, narrowed lumbar interpedicular distances, squared ilia, and macrocephaly without ACH's craniofacial features .
- Skeletal surveys are essential, as no single feature is pathognomonic; delayed diagnosis may occur in adulthood.
- Differentiation from ACH requires evaluation of lumbar spine, pelvis, and hand morphology.
- Regular imaging monitors complications like spinal stenosis and osteoarthritis, especially in adults.

ETHICAL COMPLIANCE

This pictorial review uses radiographic images (image1.jpeg, image2.jpeg, image3.jpeg, image4.jpeg) and a schematic diagram to illustrate the radiological findings of hypochondroplasia in a single 44-year-old male patient, with a clinical photograph placeholder. The radiographic images were de-identified to ensure complete anonymization, and their use complies with institutional ethical guidelines, including informed consent obtained from the patient or authorized representative. Figure 6 is an original schematic creation. Figure 1 (clinical photograph) is a placeholder pending submission of a de-identified image with consent. This study complies with the Declaration of Helsinki and relevant ethical standards for medical publication.

DISCLOSURES

➢ **Funding**: No external funding was received for this study.

➢ **Conflicts of Interest**: The authors declare no conflicts of interest related to this manuscript. None of the authors have financial or personal relationships with organizations that could influence the work, including but not limited to pharmaceutical companies, imaging equipment manufacturers, or diagnostic services.

➢ **Author Contributions**: Dr. Mrudula R, MD: Conceptualization, manuscript drafting, figure selection. Dr. B.K.D. Prasad, MD: Table design, orthopedic perspective. Dr. Akanksh Chokkapu, MD: Radiological interpretation, critical revision. All authors approved the final manuscript and agree to be accountable for its content.

➢ **Acknowledgments**: None.

➢ **Data Availability**: De-identified radiographic images (image1.jpeg, image2.jpeg, image3.jpeg, image4.jpeg) are included; the clinical photograph (Figure 1) is a placeholder pending submission. Additional high-quality radiographic images are available on request from institutional archives, ensuring compliance with ethical standards.

REFERENCES

[1]. Bober MB, Bellus GA, Nikkel SM, et al. Hypochondroplasia. GeneReviews®. 2002 [updated 2020]. doi:10.1007/978-3-540-29676-8_1477
[2]. Sabir AH, Sheikh J, Singh A, et al. Earlier detection of hypochondroplasia: A large single-center UK case series and systematic review. Am J Med Genet A. 2021;185(1):73-82. doi:10.1002/ajmg.a.61912
[3]. Loisay L, Komla-Ebri D, Morice A, et al. Generation and characterization of a mouse model for hypochondroplasia. JCI Insight. 2023;8(16):e168051. doi:10.1172/jci.insight.168051
[4]. Evaluation of Efficacy of Long-term Growth Hormone Therapy in Patients with Hypochondroplasia. J Clin Res Pediatr Endocrinol. 2013;5(3):279-284. doi:10.4274/jcrpe.0043
[5]. Skeletal Survey & Diagnostic Yield. J Clin Res Pediatr Endocrinol. 2021;13(3):363-368. doi:10.4274/jcrpe.galenos.2021.2021.0069
[6]. The skeletal dysplasias: an overview. Indian J Radiol Imaging. 2015;25(2):100-104. doi:10.4103/0971-3026.155823
[7]. Spine radiograph in dysplasias: A pictorial essay. Indian J Radiol Imaging. 2021;31(3):755-766. doi:10.4103/ijri.IJRI_395_20
[8]. Prenatal diagnosis of achondroplasia and hypochondroplasia using three-dimensional computed tomography: a retrospective study. Quant Imaging Med Surg. 2023;13(10):6687-6695. doi:10.21037/qims-23-118
[9]. Spinal manifestations of achondroplasia. Pediatr Radiol. 2021;51(7):1109-1119. doi:10.1007/s00247-021-04987-1
[10]. Imaging of Fibroblast Growth Factor Receptor 3-related Skeletal Disorders. Radiographics. 2017;37(7):2025-2041. doi:10.1148/rg.2017170017
[11]. The radiographic manifestations of hypochondroplasia. Clin Radiol. 1979;30(1):79-85. doi:10.1016/s0009-9260(79)80051-3
[12]. Hypochondroplasia. Arch Dis Child. 1978;53(11):868-872. doi:10.1136/adc.53.11.868
[13]. Comparison of clinical-Radiological and molecular findings in hypochondroplasia. Am J Med Genet. 1998;78(1):1-5. doi:10.1002/(SICI)1096-8628(19980602)78:1<1::AID-AJMG1>3.0.CO;2-H

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25jun1833

[14]. Treatment of Varus Deformities of the Lower Limbs in Patients with Achondroplasia and Hypochondroplasia. J Clin Orthop Trauma. 2013;4(1):15-20. doi:10.1016/j.jcot.2013.01.003
[15]. Hypochondroplasia. Endocrinol Metab (Seoul). 2022;37(3):363-370. doi:10.3803/EnM.2022.1384