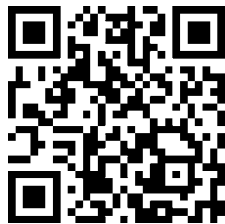


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Diagnosis of hypochondroplasia: International Delphi consensus recommendations



Scan the QR code or click [here](#)
to view the guidelines

Hypochondroplasia is a rare genetic skeletal dysplasia with diagnostic challenges

Guidelines provide a standardized framework for timely, accurate diagnosis

- Led by multidisciplinary experts from the International Achondroplasia Forum for achondroplasia and related conditions (IAF)
- Applicable across diverse healthcare settings globally

Key recommendations address:



Clinical,
radiographic, and
genetic evaluations



Family history
and inheritance



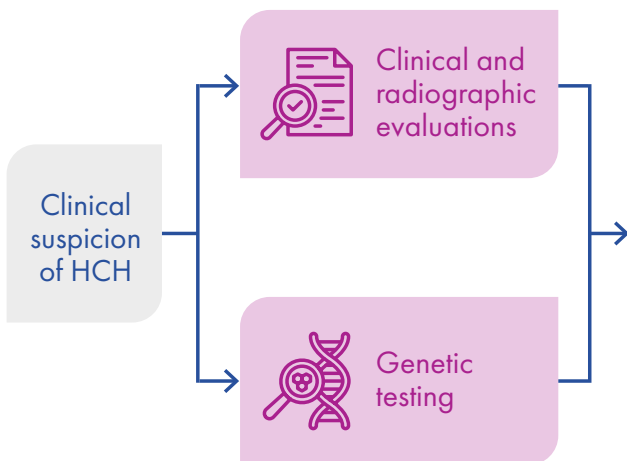
Prenatal
diagnosis

The diagnostic framework begins with
clinical suspicion of hypochondroplasia



Diagnostic framework for hypochondroplasia

Leading to three potential diagnostic outcomes



FGFR3 genetic testing result	No. of major criteria	No. of minor criteria*	Diagnosis†
Pathogenic	≥1	–	Definitive HCH
Pathogenic	0	≥3	Definitive HCH
Likely pathogenic	≥2	–	Definitive HCH
Likely pathogenic	1	≥3	Definitive HCH
Likely pathogenic	0	≥6	Suspected HCH
VUS	≥2	–	Suspected HCH
VUS	1	≥3	Suspected HCH
No variant detected	≥4	–	Suspected HCH
Genetic testing unavailable/not performed	≥4	–	Genetically unconfirmed HCH

If criteria are not met:

- Consider performing additional clinical, radiographic, and/or genetic evaluations
- Repeat investigations over time, as signs may become more apparent with age

 Recommend family screening and genetic counseling

 Investigate further and re-evaluate over time

 Prenatal screening may facilitate early identification

*'–' indicates that any number of criteria (including zero) is acceptable for that pathway

†The scenarios leading to a diagnosis of suspected HCH require the absence of a definitive diagnosis of an alternate skeletal dysplasia

FGFR3, gene encoding fibroblast growth factor receptor 3; HCH, hypochondroplasia; VUS, variant of uncertain significance

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View the published guidelines for full major and minor clinical criteria