

Recognizing red flags in children with short stature*

Children with short stature and **red flag clinical features** that may suggest hypochondroplasia should be considered for **further evaluation**, including genetic testing¹⁻⁴

Disproportionate short stature[†]

Head shape and size

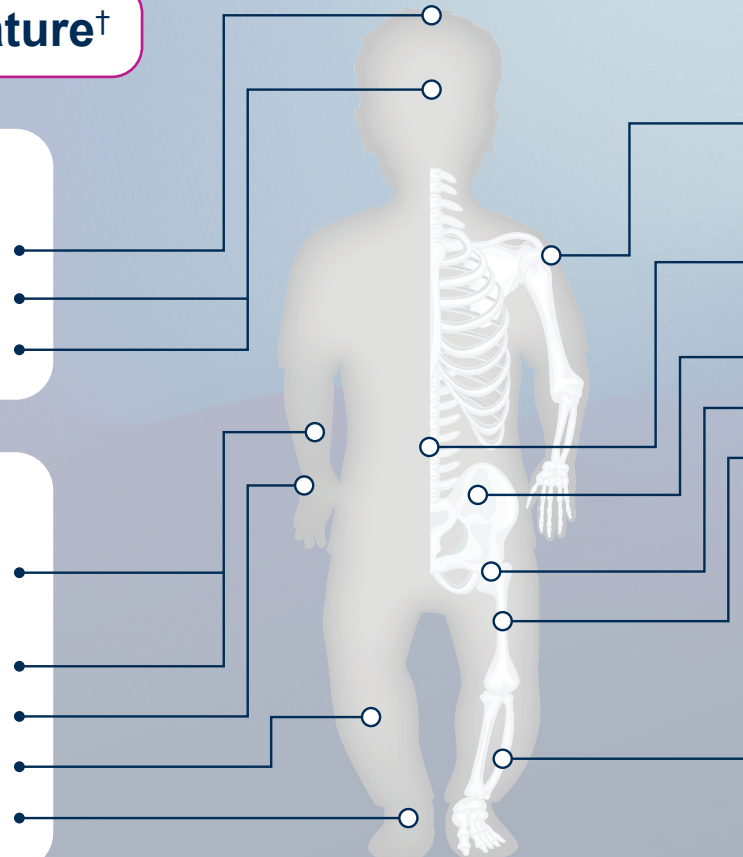
- Relative macrocephaly
- Midface hypoplasia
- Frontal bossing

Limbs

- Short arms and legs (rhizomelia/mesomelia)
- Limited arm extension
- Broad hands
- Bowed legs (or genu varum)
- Brachydactyly

Radiological signs

- Prominence of the deltoid muscular insertion on the proximal humerus
- **Progressive lumbar canal stenosis (L1–L4)**
- Small iliac bones
- Short, broad femoral neck
- Short, relatively broad long bones
- Relatively long distal fibulae



Order a genetic test at no charge⁵

Please note that no single radiologic or clinical feature is unique to hypochondroplasia and the expression of many of the diagnostic features in affected individuals is variable

*Red flags refer to clinical features that may raise suspicion for hypochondroplasia and prompt further evaluation, including consideration of genetic testing. Features shown are not intended to represent all possible manifestations; †Assessment of body proportions may help identify disproportionate short stature suggestive of skeletal dysplasia, including hypochondroplasia

1. Sharma L *et al.* *Short Stature*. In StatPearls [Internet]. 2025. Available at: <https://www.ncbi.nlm.nih.gov/books/NBK556031/>. Accessed May 2026; 2. Bober MB *et al.* *Hypochondroplasia*. In GeneReviews® [Internet]. 1999 (updated September 25, 2025). Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1477>. Accessed May 2026; 3. Sabir AH *et al.* *Am J Med Genet* 2021;185A:73–82; 4. Irving M *et al.* *Adv Ther* 2026;doi:10.1007/s12325-026-03526-2; 5. Labcorp. Discover Dysplasias Program. Available at: <https://www.invitae.com/us/sponsored-testing/discover-dysplasias>. Accessed May 2026

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