#TAKE 5 to change lives

The symptoms of **acrodysostosis** can be subtle, but recognising the signs could unlock a diagnosis that paves the way to **vital support and resources.**

Brachydactyly is often the most noticeable symptom. We've created the **HANDS** acrostic to help you remember this and other important indicators.

H

Hands and feet are abnormally small with short, stubby fingers and toes (brachydactyly)[1–3]

A

Atypical skeletal formation, including possible spinal curvature abnormalities[2]

N

Nasal structure is flatter and face is broader with widely spaced eyes[1–3]

D

Developmental delays may result in short stature and, in some cases, intellectual disability[2,3]

S

Symptoms of hormonal deficiency can appear due to specific hormonal resistance (e.g. PTH and TSH)[3]

PTH, parathyroid hormone; TSH, thyroid-stimulating hormone.

Take 5 minutes to remember these 5 symptoms and you could change lives

WHAT IS ACRODYSOSTOSIS?

Acrodysostosis is a rare genetic condition that affects bone and physical development. Due to its rarity, it is often not considered in differential diagnoses, leading to delays in diagnosis and provision of support.[1–3]

WHAT SHOULD YOU DO IF YOU NOTICE SYMPTOMS?

1. **Consider acrodysostosis**

   If a patient presents with a combination of any of the 5 symptoms described in our HANDS acrostic, think beyond more common conditions and consider acrodysostosis in your differential diagnosis.

2. **Refer to a specialist**

   Promptly refer patients to an endocrinologist or clinical geneticist for further evaluation and testing.

3. **Request genetic testing**

   If appropriate, discuss molecular testing with a specialist to confirm a diagnosis, especially if the presentation is unclear.

4. **Act early**

   Timely referral and testing can help patients access the care and support they need sooner, improving outcomes and quality of life.

#TAKE5 to change lives

acrodysostosis support and research

References: 1. Genetic and Rare Diseases (GARD) Information Center. Acrodysostosis. Accessed January 2025. https://rarediseases.info.nih.gov/diseases/5724/acrodysostosis. **2.** National Organization for Rare Disorders (NORD). Acrodysostosis. Accessed January 2025. https://rarediseases.org/rare-diseases/acrodysostosis/. **3.** Orphanet. Acrodysostosis. Accessed January 2025. https:/www.orpha.net/en/disease/detail/950.