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SHORT ABSTRACT

Helpful Signs in the Imaging Diagnosis of Hereditary Musculoskeletal Disease

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Keywords: Hereditary Musculoskeletal Disorders; Dysplasia; Radiography

Learning Objective

To review and illustrate the different signs that may be useful in the imaging diagnosis of syndromes, dysplasia and other Hereditary Musculoskeletal Disorders.

Background

Identification of Hereditary Musculoskeletal Disorders is complicated because most diseases are rare and there are many diseases to memorize [1, 2].

In addition to clinical examination by a specialized pediatrician, radiography remains a very useful tool for initial identification of this group of disorders, as this examination will further guide the clinician for requesting specific and often expensive genetic tests.

Because they are easy to remember and to teach, some signs are highly valuable in the correct radiological diagnosis of genetic bone disease.

Imaging Findings

Signs can be divided into five main groups of which many examples will be shown during this lecture:

1. **Radiodensity of the skeleton**
   a. Marble bones and Rugger-Jersey sign in osteopetrosis
   b. Candle-wax dripping in melorheostosis
   c. Striation in osteopathia striata
2. **Overall morphology of the skeleton**
   a. S-sign, wedging in congenital spine deformities
   b. Accordion shaped bones in osteogenesis imperfecta (**Figure 1**)
   c. Bone twisting
3. **Specific morphology of part of the skeleton**
   a. Bell-shaped chest deformity
   b. Spade hand in storage diseases
   c. Trident hand in achondroplasia
   d. Skull deformities such as cloverleaf or copper beaten skull

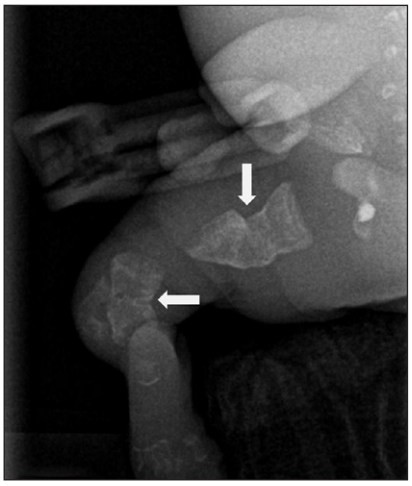

Figure 1: Accordion sign in osteogenesis imperfecta type II in a 24-month-old fetus. Plain radiograph of the right lower leg showing marked shortening of the long bones and accordion-like deformity.

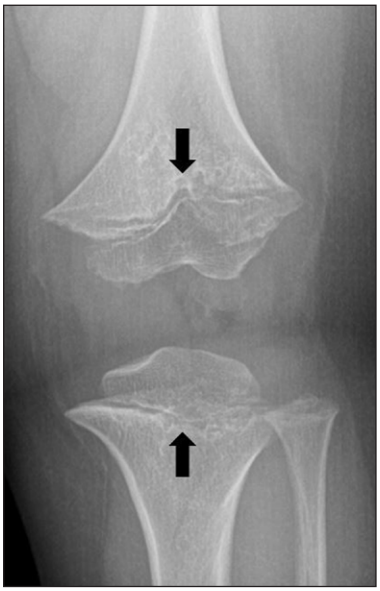

Figure 2: Chevron logo in achondroplasia. Plain radiograph of the right knee showing central depression of the metaphysis of the distal femur and the proximal tibia with focal protrusion of the adjacent epiphysis (black arrows). This resembles the Chevron logo sign.

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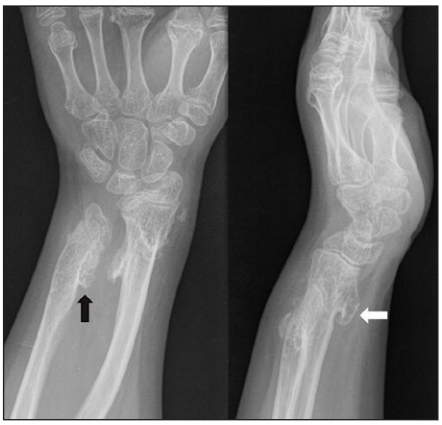

Figure 3: Cauliflower, coathanger, trumpet-shaped deformities in a patient with Hereditary Exostosis Syndrome. Plain radiograph of the left wrist showing shortening of the ulna, pseudo-Madelung deformity of the wrist and multiple exostosis. The exostosis on the distal radius has a long stalk and therefore resembles a coathanger (white arrow), whereas the exostosis on the ulna has a broad insertion on the cortex resembling a mushroom or cauliflower (black arrow).

   e. Champagne glass deformity of the pelvis in achondroplasia
   f. Slanting of joints in multiple epiphyseal dysplasia
4. **Size and shape of the individual bones**
   a. Handle bar clavicles and French telephone horns in thanatophoric dysplasia
   b. Bullet-shaped phalanges in storage diseases
5. **Size and shape of segments of bones**
   a. Cone-shaped epiphyses
   b. Pencil-shaped epiphysis in pyknodysostosis
   c. Mushroom epiphyses in epiphyseal dysplasia
   d. Cupping in metaphyseal dysplasia
   e. Chevron logo in achondroplasia (**Figure 2**)
   f. Scalloping, beaking, bullet-shaped or H-shaped vertebral bodies
   g. Squaring of iliac wings
   h. Cauliflower, coathanger, trumpet or Bayonet deformities (**Figure 3**) sign in Hereditary Exostosis Syndrome or Leri-Weill's disease

Conclusion

Systematic analysis for radiological signs may be helpful to unravel many Hereditary Musculoskeletal Disorders. Many signs are of particular didactical value as they are easy to teach and to remember.

Competing Interests

The authors have no competing interests to declare.

References

1. **Lachman, RS.** Taybi and Lachman's Radiology of Syndromes, Metabolic Disorders and Skeletal Dysplasias, 5th Edition. Mosby, St Louis; 2007.
2. **Vanhoenacker, FM, Van Hul, W, Gielen, J** and **De Schepper, AM.** Congenital skeletal abnormalities: an introduction to the radiological semiology. *Eur J Radiol*. 2001 Dec; 40(3): 168–83. DOI: https://doi.org/10.1016/S0720-048X(01)00398-9

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