SHORT ABSTRACT

Helpful Signs in the Imaging Diagnosis of Hereditary Musculoskeletal Disease

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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.
5. **Size and shape of segments of bones**
   a. Cone-shaped epiphyses
   b. Pencil-shaped epiphysis in pyknodysonostosis
   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**