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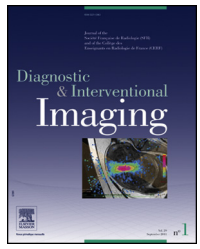

REVIEW /*Musculoskeletal imaging*

Isolated and syndromic brachydactylies: Diagnostic value of hand X-rays

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Abstract Brachydactyly, or shortening of the digits, is due to the abnormal development of phalanges, metacarpals and/or metatarsals. This congenital malformation is common, easily detectable clinically but often requires additional radiological exploration. Radiographs are essential to characterize the type of brachydactyly and to show the location of the bone shortening, as well as any associated malformation. This article reviews the radiological findings for isolated brachydactylies (according to the types classified by Bell, and Temtamy and McKusick) and for brachydactylies that are part of complex multisystem malformation syndromes. If warranted by the clinical and radiological examinations, a genetic analysis (molecular and/or cytogenetic) can confirm the etiologic diagnosis.

Brachydactyly is a congenital abnormality, which is characterized by the absence or rudimentary development of metacarpals, metatarsals, and/or phalanges. Clinically, patients present with shortened hands or feet. A postero-anterior view of the hands and feet by standard radiography is the first-line investigation to analyze the bones involved. Indeed, the analysis of the affected digit as well as the topography of the shortened bone within the digit, allows the radiologist to determine the classification of the

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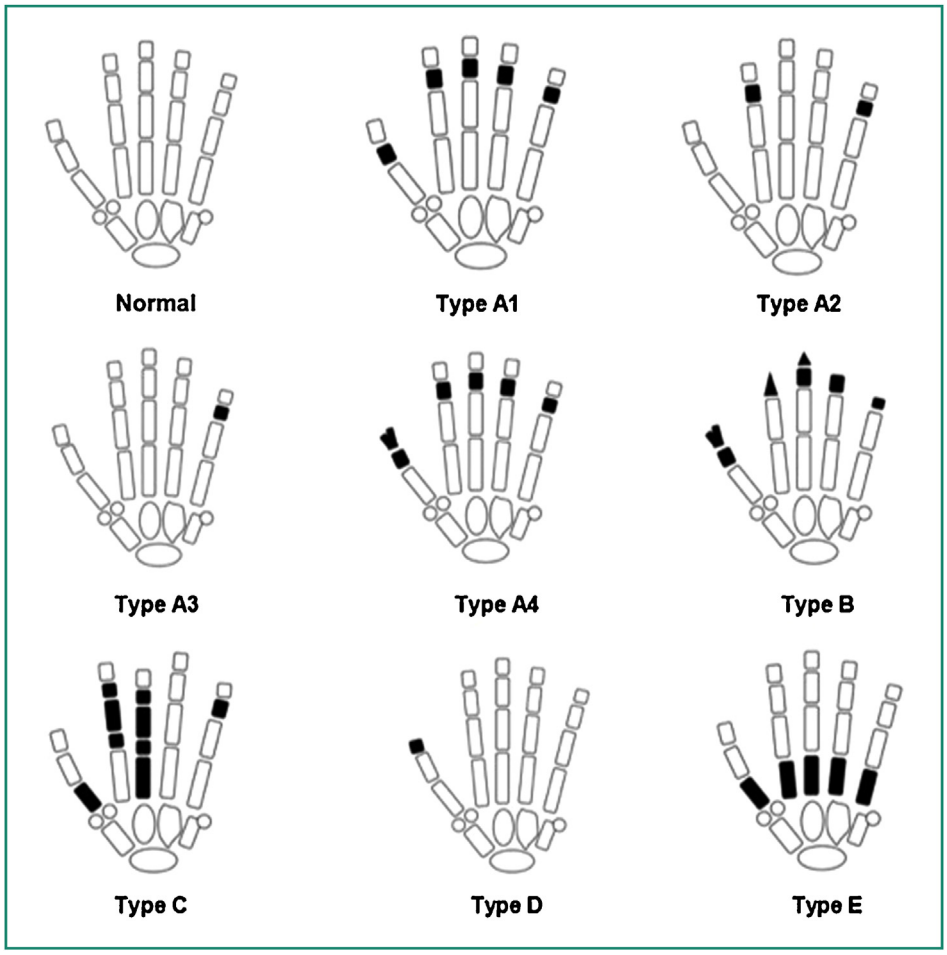

Figure 1. Classification of isolated brachydactylies. Temtamy's classification, 1978.

brachydactyly and whether the condition is to be diagnosed as syndromic. The abnormal shortness of phalanges is called brachyphalangia. Depending on the phalanx involved, there are three different types of brachyphalangia: brachybasophalangia (proximal phalanges), brachymesophalangia (middle phalanges) and brachytelephalangia (distal phalanges). Brachymetacarpia and brachymetatarsia are the abnormal shortness of the metacarpal and metatarsal bones; while the term clinodactyly refers to the lateral deviation of a phalanx, usually the distal phalanx.

A test (molecular or cytogenetic test, or both) to detect a genetic abnormality will further establish the etiological diagnosis. Brachydactyly may be an isolated condition or part of a complex malformation syndrome.

Isolated brachydactyly

Isolated brachydactyly is one of the ten categories of hand malformations described by Temtamy and McKusick. Bell initiated a classification in 1951 [1] and Temtamy and McKusick further completed it in 1978 (Fig. 1) [2]. This classification consists of five individualized types A to E, and four subgroups A1 to A4. These malformations are rare, except for types A3 and D, more frequent, for which the prevalence reaches 2% [2].

Type A

Brachydactyly type A is characterized by shortened middle phalanges. Depending on the affected digit, brachydactyly type A is subdivided into four subtypes, A1 to A4.

Type A1

Type A1 is shortened or undeveloped middle phalanx in all digits, as well as proximal phalanx of thumb (or big toe). Second and fifth fingers are the most frequently affected. Hand X-rays show short or absent middle phalanges, short distal phalanges, and sometimes fusion of distal and middle phalanges (Fig. 2) [3]. Metacarpals are often short with broad epiphysis [4]. Brachydactyly type A1 is inherited in an autosomal dominant pattern and is caused by mutations in gene *IHH* (Indian Hedgehog) located on chromosome 2 at 2q35-36 [5].

Type A2

Type A2 is absent or shortened middle phalanx of the index finger, or more rarely 5th digit [2]. Diagnosis is confirmed on