Identification of congenital skeletal abnormalities is complex because of the large variety of individual syndromes and dysplasias that are often difficult to remember. Although a correct diagnosis relies on a combination of clinical, radiologic, and genetic tests, imaging plays an important role in selecting those patients who should be referred for further genetic counseling and expensive genetic tests. In addition to information derived from radiologic analysis of other skeletal elements, radiographs of the hand and wrist may provide particular useful information. In the first part of this article, we provide a guide for a systematic radiologic analysis of the hand and wrist bones that may help characterize congenital and developmental diseases. Special attention is given to the use of correct terminology. In the second part, we discuss typical examples of congenital and developmental diseases involving the hand and wrist, with an emphasis on skeletal dysplasias.

Radiologic Semiology and Terminology

Radiologic analysis is based on the evaluation of cardinal and additional criteria, summarized in Table 1 and further illustrated here.

Cardinal Criteria

Overall Morphology

The overall morphology of the hand is the first parameter to examine. In achondroplasia a characteristic trident morphology of the hand and wrist is seen (Fig. 1). Storage diseases such as mucopolysaccharidoses causing dysostosis multiplex congenita may result in a very plump hand resembling a spade (hence the name “spade hand”) (Fig. 2).
Length of Bones
The next step is to evaluate the length of the individual bones of the hand, looking for shortening of various phalanges, thumb, and metacarpals.

"Brachydactyly: is the general term for short digits that can be subdivided in eight different subtypes, originally described by Bell (A1–4, B, C, D, and E), although it is not always possible to classify every individual case of brachydactyly with this classification (Fig. 3).

"Brachymetacarpalia" refers to a more specific term used to designate short metacarpals, whereas brachytelephalangy and brachymesophalangy are used to indicate shortening of the distal and middle phalanges, respectively. Shortening of metacarpal 4 (and 5), causing a positive metacarpal sign (Archibald’s sign) is seen in a variety of disorders including Turner’s syndrome, pseudohypoparathyroidism, and pseudo-pseudohypoparathyroidism. This sign is positive when a line drawn along the heads of the fourth and fifth metacarpals intersects the head of the third metacarpal (Fig. 4). Shortening of the fourth metacarpal may occur as a normal variant as well.4,5

In many conditions, overlapping signs of brachymetacarpalia and brachydactyly are seen.3

Arachnodactyly are elongated and slender tubular bones of the hand and may be seen in inherited connective tissue disorders including Marfan’s syndrome (Fig. 5), Beals syndrome, homocystinuria, and Ehlers-Danlos syndrome.3

Number of Bones
Abnormalities in the number of bones are referred to as oligodactyly (Fig. 6), in case of lack of fingers, and polydactyly, if there are too many fingers (Fig. 7).

Based on the location of the absent digits, the terminology “radial ray” (Fig. 6), “ulnar ray” syndrome, central ray (mesoaxial) deficiency, or terminal transverse defects is used.7

Ectrodactyly, also known as split, cleft, or lobster hand, is a descriptive term previously used to designate a central ray deficiency. The name “ectrodactyly” is derived from the Greek ekstra (“abortion”) and daktylos (“finger”) reflecting the deficiency or absence of one or more central digits of the hand (Fig. S1). It may also involve the foot.

Amniotic band syndrome is a condition believed to occur as a result of constriction bands in utero. The presence of ringlike depressions in the skin, which are often indirectly visible on radiographs, is the clue to differentiate amniotic band syndrome from transverse defects.
Complete absence of the hand due to a transverse deficiency is known as acheiria. A supernumerary finger can be either preaxial (on the radial side), postaxial (on the ulnar side) (Fig. 7), mesoaxial (middle finger), or complex (comprising combined abnormalities). In case of mesoaxial polydactyly, there is often a Y-shaped duplication of the third metacarpal. Polydactyly can occur in association with short ribs, in a number of polydactyly-short rib syndromes (Fig. 7). Ulnar dimelia is very rare and characterized by duplication of the ulna, absence of the radius, radial carpal bones and fingers, and postaxial polydactyly with midline symmetry, hence the name "mirror hand" (Fig. 8).

A triphalangeal thumb is characterized by an elongated thumb comprising three rather than the usual two phalanges and can present as a sporadic occurrence or in association with other inherited hand and foot abnormalities (Fig. S2).

Fusion of Bones
Fusion occurring between two or more (adjacent) fingers is called syndactyly (Fig. 9), whereas symphalangism (Fig. 10) refers to fusion between two phalanges within the same ray. Because symphalangism represents a failure in segmentation, the term "fusion" is a misnomer. Syndactyly can be...

**Fig. 2** Mucopolysaccharidoses. Radiograph of the right hand showing a “spade hand” in Hurler’s syndrome. Note also flexion deformity of the fingers at the level of the interphalangeal joints, most pronounced at the distal interphalangeal joint of the fifth finger. There is a bullet-shaped deformity of the proximal and middle phalanges and pointing of the proximal ends of metacarpals 1 to 4.

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**Fig. 3** Bell’s classification of brachydactyly.

**Fig. 4** Pseudohypoparathyroidism. Radiographs of (a) the right hand and (b) thumb showing shortening of metacarpal 1 and particularly of metacarpals 4 and 5 (positive metacarpal sign) and soft tissue calcifications at the dorsal aspect of the proximal phalanx of the thumb.
membranous, bony (►Fig. 9), or combined and located either preaxial (on the radial side), postaxial (on the ulnar side), mesoaxial (middle finger), or associated with polydactyly (sympolydactyly).

Symphalangism can be subdivided based on the location as proximal (at the proximal interphalangeal joint), distal (at the distal interphalangeal joint), or variable (distally and proximally located within the same patient) (►Fig. 10). Symphalangism may be completely bony or fibrous.

Fusion between carpal (or tarsal) bones is a frequent finding (►Fig. 11 and ►Fig. S3) that is usually isolated but can be associated with several syndromes.10,11 Similar to symphalangism, the abnormality represents a failure in segmentation, and therefore the term “fusion” or “coalition” is a misnomer.8 Isolated coalitions usually involve two adjacent carpal bones within the same row, whereas multiple coalitions extending across the carpal rows may be associated with congenital syndromes.8,12

Shape of Bones
Changes in morphology may involve the phalanges, the carpal, metacarpals, and distal radius and ulna. The phalanges may have a bullet shape in a variety of skeletal dysplasias including achondroplasia and dysostosis multiplex congenita13 (►Figs. 1 and 2).

“Acro-osteolysis” is the term used for distal phalangeal resorption. This is usually due to acquired causes such as frostbite, scleroderma, psoriasis, and Raynaud’s phenomenon, but congenital disorders such as pyknodystosis14 (►Fig. 12) or progeria (►Fig. 13) or other progeroid syn-

Fig. 5  Arachnodactyly in Marfan’s disease. Radiographs of the left hand show long and slender metacarpals and phalanges.

Fig. 6  Radial ray syndrome. Radiographs showing bilateral absence of the radius and the thumb (oligodactyly).

dromes may also cause acro-osteolysis or pointing of the distal phalanges (►Fig. S4).

The most frequent abnormality regarding the morphology of the individual bones consist of cone-shaped epiphyses of the phalanges, either occurring as an isolated finding (►Fig. 14) or as part of an inheritable disorder (►Fig. 7).

A delta phalanx or longitudinal epiphyseal bracket is caused by C-shaped secondary bone growth in an aberrant epiphysis extending along one side of the diaphysis. This leads to asymmetric longitudinal growth of the diaphysis, resulting in a rhomboid or triangular shape of the phalanges.3,15 It most commonly affects the proximal phalanx of the thumb (►Fig. 15) or the middle phalanx of the fifth finger.16,17

Angel-shaped phalanges, resembling little angels of a Christmas tree, are unusual and may be seen in angel-shaped phalangeal-epiphyseal dysplasia, a rare autosomal dominant inherited disorder characterized by delayed bone age, abnormal dentition, hyperextensibility of the interphalangeal joints, and premature coxartrosis.3

Pseudo-epiphyses are “epiphyses” located at an abnormal site such as the base of the second metacarpal. They may be seen as a normal variant, particularly at the base of the second and fifth finger (►Fig. S5), or occur in cleidocranial dysplasia, brachydactyly C, chondro-ectodermal dysplasia, Fanconi’s
anemia, Kniest disease, Larsen’s syndrome, otopalatodigital syndrome, and chromosomal abnormalities.

Metaphyseal changes consist of splaying, cupping, widening, and irregularity (Fig. 16).

Other morphological changes may involve the carpal bones and consist of smallness, scalloping, and irregularity (Fig. S6).

At the level of the distal radius and ulna, one has to look for an abnormal carpal angle that should range between 130 and 137 degrees. A decreased carpal angle is associated with Turner’s syndrome, Leri-Weill dyschondrosteosis, dysostosis multiplex congenita, and Madelung’s deformity or pseudo-Madelung’s due to hereditary multiple exostosis (HME) syndrome. The carpal angle may also be enlarged in Down syndrome and a number of dysplasias.

**Additional Criteria**

**Abnormal Bone Age**

The bone age is delayed in many congenital diseases or dysplasias, although this is a nonspecific finding that may present in many other disorders.
Abnormalities in Mineralization, Either Sclerosis or Demineralization

Ivory epiphyses of the phalanges are usually a normal variant, but they may be part of a variety of malformation syndromes and dysplasias, such as trichorhinophalangeal syndrome, Cockayne’s syndrome, and epiphyseal dysplasia. In contrast to normal variants, ivory epiphyses associated with congenital disorders are usually multiple and occur in combination with other anomalies. Normal variants typically involve the distal phalanges of the second to fifth finger or the middle phalanx of the fifth finger. They disappear after closure of the growth plate. Sclerosis, fragmentation, and irregularity of the epiphyses of the proximal interphalangeal and distal interphalangeal joints can also occur in Thiemann’s syndrome,

Fig. 11 Carpal coalition. Note failure of segmentation between the lunate and the triquetrum.

Fig. 12 Pyknodysostosis. Radiograph of the left hand showing osteolysis of the distal phalanx of the second finger and subtle irregularity of the tuft of the distal phalanges of the thumb, third, and fourth finger.

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Fig. 13 Progeria. Radiographs of the right hand showing subtle acroosteolysis at the terminal phalanges of the second to fifth finger. Subtle changes are often difficult to distinguish from the normal blunted appearance of the tufts of the distal phalanges in young children.

Fig. 14 Cone-shaped epiphyses as incidental findings. Radiograph of the left hand showing cone-shaped epiphyses at the middle phalanx of the second and fifth finger.
an autosomal dominant inherited familial osteoarthropathy with osteonecrosis.\textsuperscript{19,20}

Sclerosing bone dysplasias, affecting the intramembranous bone formation and remodeling, are responsible for cortical thickening.\textsuperscript{21} The cortical thickening may be caused either by a disturbance in the periosteal bone formation and apposition or due to a defective endosteal resorption. According to the involved mechanism, the diaphysis may be broadened (increased periosteal apposition), the medullary cavity may be narrowed (defective endosteal resorption), or a combination of these mechanisms. Van Buchem’s disease has a predominant defective endosteal resorption (\textsuperscript{-}Fig. 17), whereas in Camurati-Engelmann disease, disturbance of the periosteal apposition is the main finding.

\textbf{Fig. 15} Delta phalanx. Note a rhomboid shape of the proximal phalanx of the thumb.

\textbf{Fig. 16} Metaphyseal changes in hereditary rickets (X-linked hypophosphatemia). Radiograph of the wrist showing osteopenia with coarse trabeculation and widening of the growth plate of the radius, irregularity of the growth plate at the metaphyseal side, and cupping of the metaphysis of the distal ulna.

\textbf{Fig. 17} Van Buchem’s disease. Radiograph of left hand showing overall increased density of the radius, ulna, metacarpals, and carpal bones. Note defective modeling of the tubular bones, cortical hyperostosis, and narrowing of the medullary cavity.
Joint Configuration
Flexion deformities of the joints of the hands may be seen in dysostosis multiplex congenita (Fig. 2).
Positional abnormalities of the fingers (and toes) can be due to an abnormal form of a phalanx or mechanical intrauterine stress. Overlapping fingers are seen in some chromosomal abnormalities (trisomy 18).

Clinodactyly consists of a radial deviation of a finger, especially the fifth finger. It usually results from an abnormal shape and shortening of the middle phalanx (Fig. 4b). It can be isolated or part of a variety of malformation syndromes.3

Camptodactyly consists of a fixed flexion deformity of the proximal interphalangeal joints, most commonly involving the fifth finger. Most cases are sporadic, but a genetic etiology with autosomal dominant inheritance may occur.3 Radiographs can confirm the flexion deformity on the lateral view but fail to demonstrate underlying soft tissues abnormalities. Ultrasound and magnetic resonance imaging (MRI) may demonstrate aberrant lumbrical insertion and decreased lumbrical size in the case of camptodactyly.22

Kirner's deformity is a combination of camptodactyly and clinodactyly of the fifth distal phalanx.8

A hitchhiker thumb is a characteristic finding seen in diastrophic dysplasia but may be seen in Rubinstein-Taybi syndrome (Fig. 18) and Pfeiffer's syndrome as well.

Calcifications
Stippled soft tissue calcifications are seen in chondrodysplasia punctata, Zellweger's syndrome, and Smith-Lemli-Opitz syndrome.

Phleboliths are characteristic for soft tissue hemangiomas and may indicate Maffucci's syndrome if combined with enchondromas. Scleroderma-like soft tissue calcifications may be encountered in Albright's osteodystrophy (Fig. 4b).

Assisted Bone and Soft Tissue Tumors
Cartilaginous exostoses are the hallmark of HME syndrome. Ollier's disease is characterized by multiple enchondromas, whereas in Maffucci's syndrome, enchondromas are associated with soft tissue hemangiomas.

Soft Tissue Findings
Macrodactyly refers to enlargement of one or more digits and may be associated with macrodystrophia lipomatosa (Fig. S8). Proteus syndrome, neurofibromatosis, hemangiomas, or lymphangiomatous tumors of the hands or feet. In summary, in addition to correct evaluation of other skeletal abnormalities, meticulous analysis of radiologic features of the hand and wrist may provide important clues to correct identification of congenital diseases of the skeleton. To achieve a more specific diagnosis, we refer to textbooks,15 lists of gamuts,5 review articles23,24 and Internet databanks on the subject that can be of particular help for the radiologist.25 The radiologist should be aware of clinical presentation and family history data.14 Ideally, the radiologic findings should be discussed with the referring pediatrician or geneticist.26

Whereas the hand is often involved in many congenital diseases, the absence of hand abnormalities may be diagnostic as well. A typical example is spondyloepiphysial dysplasia, in which the hand bones are typically normal, despite striking dysplastic changes that are present in the spine and epiphyses of the large bones.

Examples of Congenital and Developmental Diseases Involving Hand and Wrist
Achondroplasia
Achondroplasia is the most frequent skeletal dysplasia in the general population. It has an autosomal dominant inheritance and is caused by a disturbance in endochondral ossification due to FGFR3 mutation.

Clinically, patients have a short stature with predominant shortening of the proximal segments (humeri and femora), also known as a rhizomelic form of dwarfism.

In addition to bone shortening, radiographic features include disproportionately enlarged head, spinal stenosis, narrowing of the foramen magnum and thoracolumbar kyphosis, and lumbosacral lordosis.

Radiographs of the hand may show a trident hand,13 consisting of divergence of the middle three fingers at the level of the proximal interphalangeal joints, due to an inability to oppose the middle and the ring finger (Fig. 1). The middle three fingers are of equal length,13 often more obvious on clinical inspection than on radiographs. The phalanges are shortened and may have a bullet-shaped appearance (Fig. 1).27

Thanatophoric Dysplasia
Thanatophoric dysplasia is the most lethal neonatal skeletal dysplasia.28 The term “thanatophoric” is derived from the Greek word thanatos, meaning “death bearing.” Death occurs very soon after birth because of respiratory distress due to a small thoracic cage. As in achondroplasia, there is
Diastrophic Dysplasia
Diastrophic dysplasia, also called atypical achondroplasia with clubbed hands and feet, is a dysplasia due to disturbance of the endochondral bone formation. It is inherited in an autosomal recessive way because of a mutation in the DTDST (diastrophic dysplasia sulfate transporter) gene. The term “diastrophic” is derived from the Greek word diastrophism that refers to the geologic process of bending of the earth’s crust by which mountains, continents, and ocean basins are formed.25

Radiologically, the tubular bones are short, thick, and clubbed with delay in ossification and fragmentation of the epiphyses, multiple joint dislocations, and progressive scoliosis and kyphosis. Radiographs of the hand show a typical hitchhiker’s thumb with shortening and deformity of the first metacarpal with a proximally located thumb (Fig. 19).5,15,25

Chondroectodermal Dysplasia
Chondroectodermal dysplasia, also known as Ellis-van Creveld syndrome, is a skeletal ciliopathy, with an autosomal recessive inheritance due to a mutation in the EVS or EVC2 gene.29

Radiologically, the ribs are short, the acetabuli have a trident shape, and the limbs are shortened with bulbous ends. Particularly the proximal ulnae and distal radii are bulbous, whereas the distal ulnae and proximal radii are narrow, resulting in a “chicken drumstick” appearance.29

The spine and skull are normal.6,29 Radiographs of the hands show brachydactyly, cone-shaped epiphyses of the (middle) phalanges 2 through 5, and postaxial and/or mesoaxial polydactyly of the hands.6 Hypoplasia of the distal phalanges and capitâte-hamate fusion are other characteristic findings29,30 (Fig. 19).

Albright Hereditary Osteodystrophy
Albright hereditary osteodystrophy consists either of pseudo-hypoparathyroidism or pseudo-pseudohypoparathyroidism. Radiologically, soft tissue calcifications and brachydactyly type E of the hands (and feet) are seen, with a positive metacarpal sign (Fig. 4).

Pyknodysostosis
Pyknodysostosis is a sclerosing bone dysplasia due to defective endochondral bone formation affecting primary spongiosa. Its inheritance is autosomal recessive due to mutations in the CSTK gene encoding cathepsin K. Clinically, patients have a small stature. Radiologically, there is sclerosis of the tubular bones with preservation of the corticomedullary differentiation. Bone fragility is increased, and there is decreased pneumatization of the paranasal sinuses and delayed suture closure. Acro-osteolysis or pencil-like pointing of the distal phalanges of the hand is a characteristic finding (Fig. 12).31

Osteopetrosis
Osteopetrosis is a sclerosing bone dysplasia due to defective endochondral bone formation affecting the primary spongiosa. The inheritance and phenotype is heterogeneous.31

Two basic adult phenotypes of osteopetrosis are distinguished. In type I, uniform sclerosis is found in the skull, spine, and long bones, whereas endobones (“bone-in-bone”) and rugger-jersey spine are present in type II.21,32 The bones of the hands may be involved as well.

Osteopoikilosis
Osteopoikilosis is a disturbance of endochondral ossification that consists of an error in resorption or remodeling of secondary spongiosa, resulting in focal multiple bone islands at the end of tubular bones and at the carpal and tarsal bones. These sclerotic foci have typical peripheral thorny radiations, blending with the trabeculae of the adjacent secondary spongiosa (Fig. 20).

Melorheostosis
Melorheostosis is a mixed sclerosing bone dysplasia due to a disturbance of intramembranous and, to a lesser degree, endochondral ossification.21 It is not inherited and believed
to occur as a result of a defect in the early embryological stage, where cartilage-forming cells migrate from the sclerotomes to the limb buds. Radiologically, there is typical unilateral involvement (hemimelic) “flowing” hyperostosis of the cortex of the bones included resembling wax dripping down one side of a candle. Soft tissue involvement with a “myositis ossificans” and an “ostema-like” pattern may occur as well. There are several reports on involvement of the bones of the hand.

Progeria
Progeria is a rare autosomal dominant disorder due to a new mutation characterized by accelerated aging at a very young age. The word “progeria” comes from the Greek words pro meaning “before” or “premature,” and geras meaning “old age.” Although the term progeria refers to all diseases characterized by premature aging symptoms, it is often specifically used to designate Hutchinson-Gilford progeria syndrome. Radiographs of the hand may show scleroderma-like lesions including calcifications and acro-osteolysis (Fig. 13).

Hereditary Rickets
Hereditary rickets are a group of disorders with a heterogeneous inheritance. Radiologically, they do not differ from secondary rickets, showing demineralization with coarsened trabeculae, bowing of the long bones, poorly ossified epiphyses, widening of the growth plates, metaphyseal cupping, and fraying and flaring (Fig. 16).

Hereditary Multiple Exostosis Syndrome
HME syndrome is an autosomal dominant inherited bone disorder with an incidence of 1 in 50,000. It is a genetically heterogeneous disease, and most cases are linked to loss-of-function mutations in EXT1 or EXT2.

There is defective endochondral bone formation, resulting in multiple exostoses or osteochondromas at the juxtaepiphyseal regions of bones.

In addition, there is also a defective (re)modeling at the metaphyses of the long bones, leading to mild dwarfing, metaphyseal widening, with the formation of trumpet-shaped bones and wrist deformities ("bayonet hand"), owing to shortening of the ulna. Common osseous deformities include a pseudo-Madelung’s deformity of the wrist, with ulnar shortening and (secondary) radial bowing. Complications may occur due to local entrapment of vessel, tendon, nerve, bursitis de novo, and sarcomatous transformation.

Metachondromatosis
Metachondromatosis (MC) is a rare disorder characterized by the presence of both multiple enchondromas and osteochondromatosis-like lesions, most commonly occurring at the hands or feet. MC has an autosomal dominant mode of inheritance and is caused by loss-of-function mutations including deletions, nonsense mutations, and splice sites mutations of the PTPN11 gene (12q24). Compared with HME, no malignant transformation has been described.

Ollier’s Disease
Enchondromatosis, also known as Ollier’s disease, consists of multiple enchondromas, often affecting the hand and wrist. The estimated prevalence of Ollier’s disease is 1 in 100,000. Ollier’s disease is not inherited and has a sporadic occurrence.

There is usually an asymmetric distribution of the lesion, affecting one side of the body. Affected bones are often shortened and curved. Forearm deformities are frequently encountered similar to those observed in HME. Radiographs show multiple enchondromas (Fig. S11) and widening of the metaphysis. There is a potential risk for a malignant change to chondrosarcoma.

Maffucci’s Disease
Multiple enchondromas associated with soft tissue hemangiomatosis is known as Maffucci’s syndrome. Similar to Ollier’s disease, Maffucci’s disease is not inherited and has a sporadic occurrence. The hand and wrist are often involved. In addition to multiple enchondromas, radiographs show typical phleboliths that are characteristic for soft tissue hemangiomas.

Pachydermoperiostosis
Pachydermoperiostosis is a rare disorder with an autosomal recessive or dominant inheritance. Mutations in the HPGD gene were found in those with the autosomal recessive form.

Clinically, there is thickening of the skin of the face (pachyderma), excessive sweating (hyperhidrosis), and clubbing of the fingers. The disease starts at childhood age or at puberty, and it progresses slowly for ~10 years.
The hand may serve as an important window to the diagnosis of congenital disorders of the skeleton. We recommend scrutinizing the radiologic semiology systematically as the first step in successful characterization. In addition, every case should be discussed with the referring pediatrician and/or clinical geneticist to obtain a more specific diagnosis. Finally, the radiologist should be familiar with the main features of most common inherited bone diseases.

Conflict of Interest
None declared.

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