RadioGraphics

Obstetric US: Watch the Fetal Hands[1]

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CME FEATURE

See accompanying test at http://www.rsna.org/education/rg_cme.html

LEARNING OBJECTIVES FOR TEST 4

After reading this article and taking the test, the reader will be able to:

- Explain the importance of US examination of the fetal hands.
- Describe the most common hand malformations and the conditions with which they are associated.
- Propose an appropriate work-up for hand anomalies detected at US.

TEACHING POINTS

See last page

Hand anomalies are difficult to diagnose and are often overlooked during prenatal ultrasonography (US). The spectrum of malformations varies from subtle finger deformities to the complete amputation of limbs. Malformations of the hand can be classified, according to the predominant anomaly, among the following categories: alignment abnormalities (clenched hand, camptodactyly, clinodactyly, hypokinesia, clubhand, phocomelia), thumb anomalies, abnormal size (macrodactyly, trident hand), abnormal echogenicity (abnormal calcifications), abnormal number (polydactyly, syndactyly, ectrodactyly), and constriction band sequence. A fetal hand anomaly has important diagnostic and prognostic implications as well as functional consequences. Malformation may be isolated but often is associated with a syndrome or karyotype anomaly. Classification and characterization of the anomaly help to narrow the differential diagnosis: Some malformations (clenched hand, hitchhiker thumb) are highly suggestive of a specific diagnosis. The detection of a fetal hand malformation warrants a complete work-up, including complete fetal and cardiac US examinations, as well as genetic counseling to determine whether familial inquiry and karyotype analysis are necessary.

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Abbreviations: FADS = fetal akinesia deformation sequence, TAR = thrombocytopenia and absent radius, VACTERL = vertebral, anal, cardiac, tracheal, esophageal, renal, and limb [abnormalities]

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See the commentary by Angtuaco following this article.

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Introduction

Teaching Point

Hand anomalies encompass a large spectrum of malformations, from subtle defects of distal phalanges, almost undetectable in utero, to severe abnormalities such as clubhand, split hand, or absence of a limb. Like most other forms of skeletal dysostosis, hand anomalies are frequently missed during routine fetal ultrasonography (US) (1,2). However, a systematic examination of the fetal extremities is worth the effort, because knowledge of fetal hand anomalies can help guide the diagnosis and management of many associated conditions (3). Hand anomalies may be isolated or associated with other skeletal and nonskeletal malformations, aneuploidies, syndromes, and bone dysplasias (4,5).

A hand malformation may be fortuitously discovered during routine fetal US, but most often the sonographer will actively look for it after the identification of another major anomaly, such as intrauterine growth retardation, significant shortening of the long bones, an abnormally small thorax, or craniostenosis. In these circumstances, the discovery of a hand malformation can help to narrow the differential diagnosis. Some hand malformations are even specific enough to enable a conclusive diagnosis: The presence of hitchhiker thumb is suggestive of diastrophic dysplasia; that of a clenched hand or hands, trisomy 18; and that of trident hand, a severe form of osteochondrodysplasia (3). Last, in cases in which there is a positive family history or a previous exposure to known teratogens, a focused US examination is performed.

Teaching Point

Teaching Point

If a fetal hand anomaly is detected, the patient should be referred to a clinic that specializes in the identification and management of fetal malformations and genetic syndromes. A complete fetal work-up is necessary, including, at minimum, complete fetal and cardiac US examinations to determine the presence or absence of associated abnormalities. Repeated focused US examinations are sometimes necessary to diagnose more precisely the underlying anomalies. Genetic counseling is essential to determine whether familial inquiry and karyotype analysis are needed (4). In cases of radial clubhand, specific complementary examinations are mandatory and should include a complete fetal blood cell count to exclude thrombocytopenia and absent radius (TAR) syndrome. The possibility of Fanconi anemia should be investigated by measuring diepoxybutane-induced chromosomal breakage in fetal blood cells. Consultations with plastic and orthopedic surgeons are recommended to determine the functional prognosis.

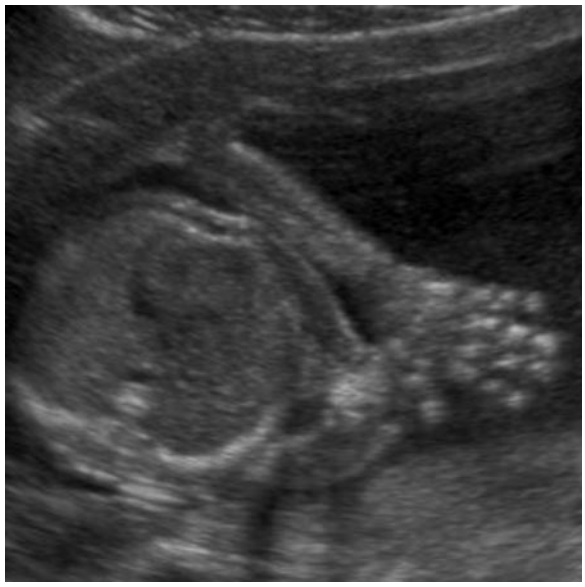
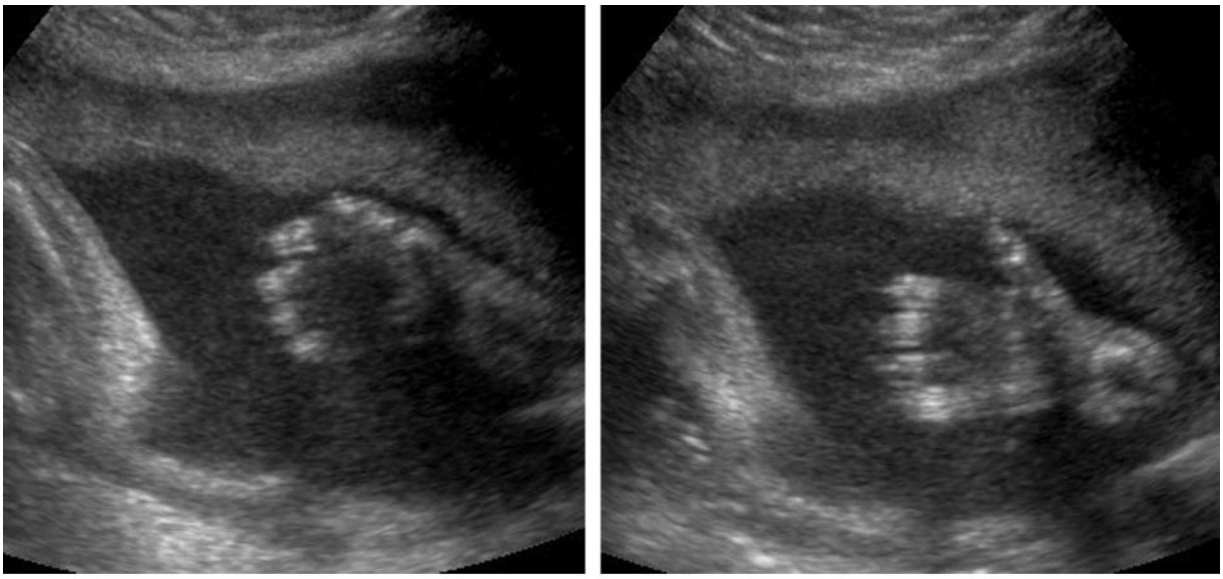


Figure 1. US image shows a normal fetal hand at 18 weeks of gestation.

Various systems exist for the classification of upper-limb malformations on the basis of anatomy, embryology, genetics, and teratology (6,7). Classification systems are useful for diagnostic and prognostic purposes. In utero, fetal hand malformations can be usefully classified as follows: *(a)* alignment abnormalities (clenched hand, camptodactyly, clinodactyly, hypokinesia, clubhand, phocomelia), *(b)* abnormal thumb, *(c)* abnormal size (macrodactyly, trident hand), *(d)* abnormal echogenicity (calcifications), *(e)* abnormal number of digits (polydactyly, syndactyly, ectrodactyly), and *(f)* constriction band sequence. In this article, the US features of these abnormalities are described with a view toward their diagnosis and management. Normal US findings are considered first, as a basis for comparison.

Normal US Findings

The best moments for US examination of the fetal hands are late in the first trimester and in the middle of the second trimester of gestation. During this time, the fetus moves frequently, and the hands are more often in an open position than they are later in pregnancy. Three-dimensional US is not mandatory but can be useful to better



a. b.

Figure 2. Fetal US images obtained at 25 weeks of gestation show a normal relaxed hand **(a)** and the same hand 1 second later **(b)**.

depict the morphologic features of the defect and the spatial relationship between the various anatomic segments or to demonstrate the anomaly more clearly (8).

Visualization of the hands is possible as soon as the long bones begin to ossify. Ossification of the humerus, radius, ulna, and phalanges begins at 11 weeks after the last menstrual cycle, and that of the metacarpals, at 12 weeks. The ossification of the carpal bones occurs after birth (9) (Figs 1, 2).

For a US examination of the fetal hands to be considered normal, the following anatomic structures must be identified, and their presence, number, size, morphology, position, and relationship must be clearly defined: the unossified hypoechogenic carpus, five hyperechoic and cylindric metacarpal bones, five independent digits of different length with three ossified phalanges (two for the thumb), and a normal radius, ulna, and humerus.

The fetal hand and wrist are normally slightly flexed, and the hand is closed at rest. Flexion and extension of the fingers and wrist must be observed. The axis between the forearm and the hand is usually neutral. No fixed position is ever normal for any joint. If possible, both hands should be examined while in an open configuration and in axial and longitudinal views.

Alignment Abnormalities

Fixed positions of the wrists or the digits are abnormal. When an alignment abnormality is fixed, the prognosis is often guarded, not only because of the functional effects but also because of the frequent association of such fixed positions with karyotype anomalies and syndromes (Table 1).

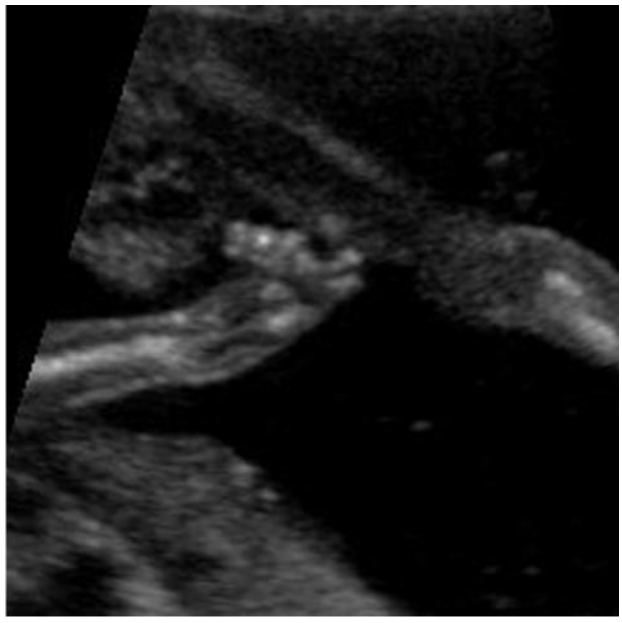
Teaching Point

Table 1
Alignment Abnormalities

US Finding	Associated Condition or Syndrome
Clenched hand	Trisomy 18, FADS
Camptodactyly	Trisomies (18, 13, 15), FADS
Clinodactyly	Trisomy 21
Clubhand	Aneuploidies, Fanconi anemia, TAR syndrome, Aase syndrome, Holt-Oram syndrome, VACTERL association
Phocomelia	Sporadic, thalidomide use, Roberts syndrome, TAR syndrome, Grebe syndrome

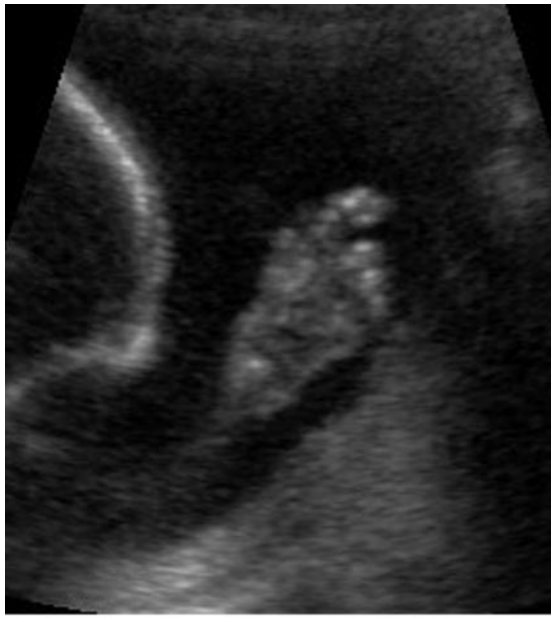
Note.—FADS = fetal akinesia deformation sequence, VACTERL = *v*ertebral, *a*nal, *c*ardiac, *t*racheal, *e*sophageal, *r*enal, and *l*imb abnormalities.

3.



4a.

Figures 3, 4. **(3)** Schematic representation of a clenched hand. **(4a)** US image shows a typical clenched hand of a fetus with trisomy 18 at 18 weeks of gestation. **(4b)** US image shows ulnar deviation of the index finger, which overlaps the other digits.



4b.

Clenched Hand

In this anomaly, the index finger overlaps a clenched fist formed by the other digits. The proximal interphalangeal articulation of the index finger is flexed and ulnarly deviated (Figs 3, 4), and the thumb is adducted. This position is constant during the examination and is strongly suggestive of trisomy 18 (5). It must be differentiated from a fixed position observed in akinesia-hypokinesia syndromes and from a temporarily closed but normal fist.

Trisomy 18, which affects one of 5000 live births, shows great phenotypic variability. Intrauterine growth retardation, craniofacial dysmorphism, congenital heart disease, omphalocele, an abnormally large cisterna magna, and abnormal extremities are frequently observed. At least 30% of affected fetuses have unilateral or bilateral limb anomalies, the most frequent of which are clenched hand and radial aplasia.

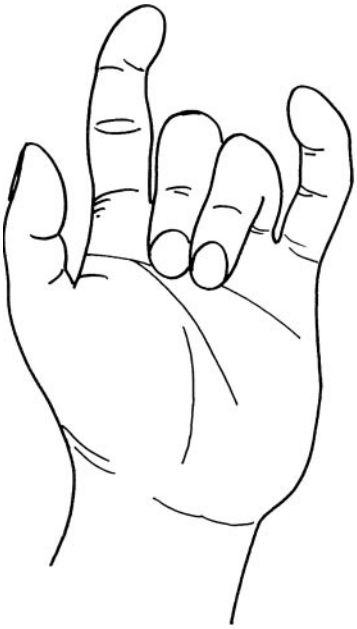
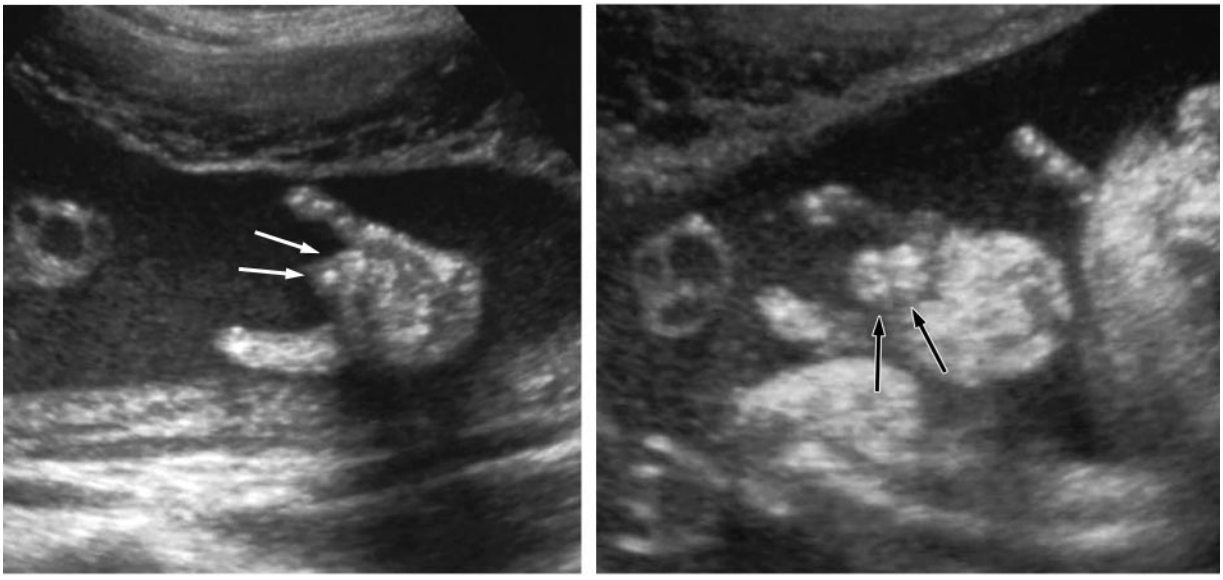


Figure 5. Focal akinesia. **(a)** Schematic representation of the anomalies observed at US in a fetus at 30 weeks of gestation. **(b)** US image shows camptodactyly, with permanent flexion of the proximal phalanges of the third and fourth fingers (arrows) and with flexion deformities of the distal interphalangeal articulations of the second and fifth fingers, which are extended. **(c)** US image, which shows the palmar aspect of the same hand, helps confirm the proximal flexion of the third and fourth fingers (arrows).

a.



b. **c.**

Camptodactyly

Camptodactyly is a flexion contracture of the proximal interphalangeal joint of the finger (Fig 5). Severe camptodactyly of multiple digits is usually associated with a karyotype anomaly (trisomy 18, 13, or 15) or a contracture syndrome. The deformation is often asymmetric, may be isolated, and may progress during infancy or childhood (6).

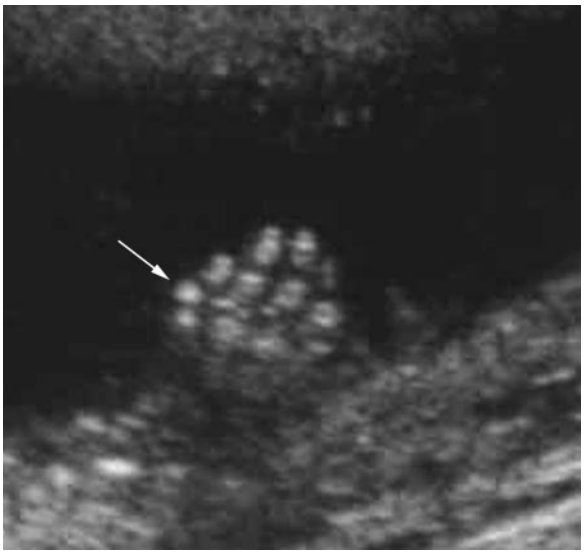


Figure 6. Clinodactyly. US image shows clinodactyly of the fifth finger in a normal fetus (arrow) at 18 weeks of gestation.

Clinodactyly

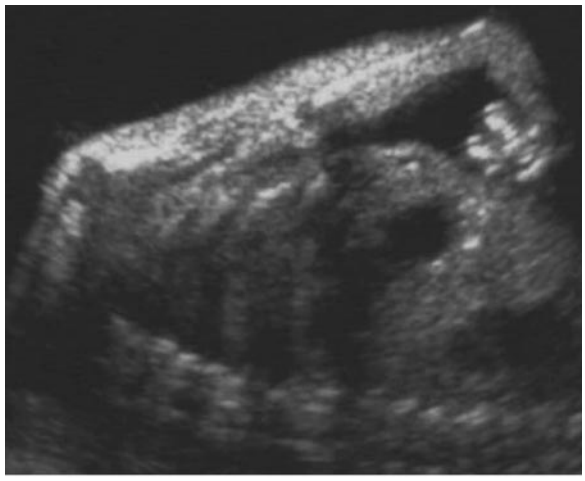
Clinodactyly is a fixed abnormal deviation of the fifth finger in the coronal or radioulnar plane. The deviation is caused by the abnormally small size of the middle phalanx, which produces a radial angulation of the distal interphalangeal joint (Fig 6).

Familial clinodactyly is usually isolated. It is inherited as an autosomal dominant trait with variable expression and incomplete penetrance (6).

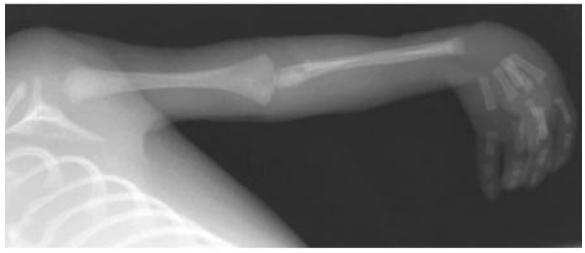
Clinodactyly has been reported in up to 60% of infants with Down syndrome. However, a high false-positive rate (18% of the normal population) makes the sign useless when it is observed as an isolated finding in a screening setting (4). Other major and minor signs suggestive of trisomy 21 (increased nuchal thickness, heart defect, ventriculomegaly, hypoplastic nasal bone) thus must be carefully sought when clinodactyly is observed at fetal US.

Hypokinesia

A prolonged decrease or absence of fetal motion may result in FADS, a group of anomalies that, in the most severe form of its expression, may include abnormal limb position, craniofacial deformation, growth restriction, polyhydramnios, lung hypoplasia, and an abnormally short umbilical cord (4). The deformities are usually symmetric, and their severity often increases distally along the involved limb. FADS is usually generalized, but



a.



b.

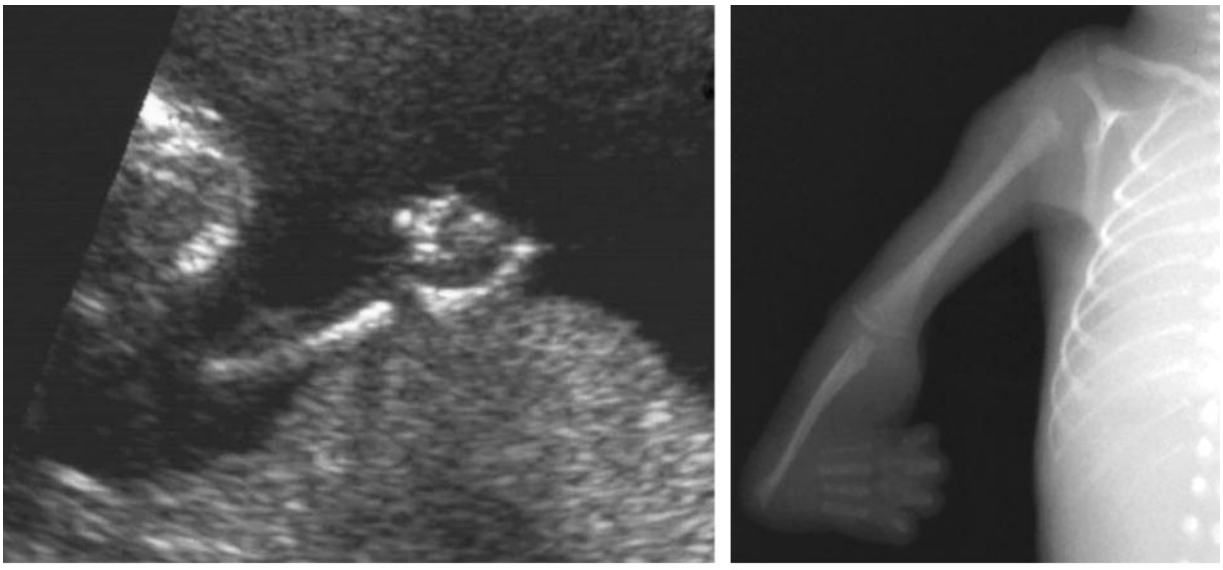
Figure 7. Diffuse akinesia. **(a)** US image in a fetus at 23 weeks of gestation shows the left wrist flexed, the hand closed, and the elbow extended, a position that remained unchanged during two successive examinations. **(b)** Corresponding radiograph of the left upper limb of the same fetus shows a clubhand and a normal radius and ulna. The other three limbs (not shown) also manifested fixed deformities.

focal anomalies do occur. Bimelic akinesia affects the lower limbs more often than it does the upper ones.

The diagnosis of FADS depends on the demonstration of fixed abnormal positions (flexion deformities, multiple camptodactylies) in association with restricted fetal movements or complete akinesia (Fig 7). Diagnosis is difficult in cases of focal akinesia.

FADS can be secondary to neurogenic diseases, myogenic anomalies, and restrictive dermopathies. Because of the various possible causes of FADS, the prediction of recurrence is difficult, and estimates vary from 0% to 25%. Generalized FADS is usually lethal because of lung hypoplasia (10,11). The functional prognosis of focal FADS is difficult to establish prenatally.

The differential diagnosis includes trisomy 18, which shares some of the craniofacial, limb, and thoracic anomalies that are associated with FADS, and fetal hypokinesia secondary to severe oligohydramnios or to uterine malformation.



a. b.

Figure 8. Radial clubhand. **(a)** US image shows severe constant malposition of the hand, secondary to the absence of one forearm bone. **(b)** Corresponding radiograph helps confirm the radial clubhand deformation secondary to radial aplasia.

Table 2
Syndromes Associated with Radial Clubhand

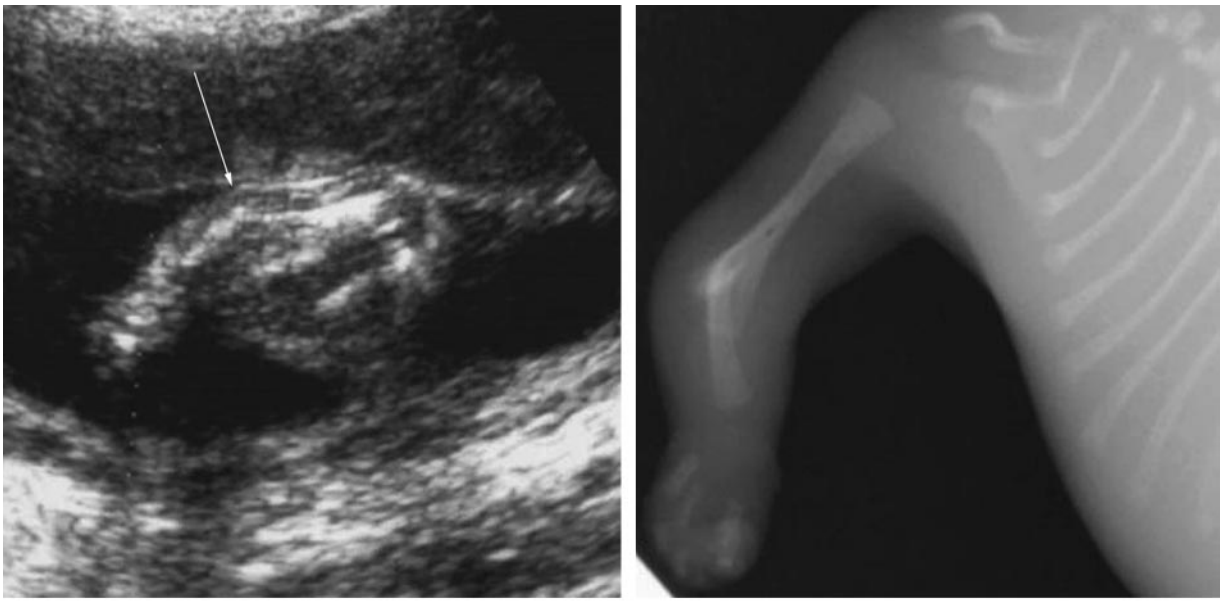
Syndrome	Characteristics
Aneuploidies	Trisomy 18, 21; chromosome 13 deletion; ring chromosome 4
Fanconi anemia	Autosomal recessive inheritance, pancytopenia secondary to bone marrow failure, radial hypoplasia, radial clubhand, absent thumb
TAR syndrome	Radial agenesis, thumb and metacarpals present, humerus and ulna may be absent, cardiac malformation (tetralogy of Fallot, ventricular septal defect) in 33% of cases
Aase syndrome	Autosomal recessive inheritance, hypoplastic anemia, radial clubhand, triphalangeal thumb, hypoplastic distal radius, cardiac defects (eg, ventricular septal defect)
Holt-Oram syndrome	Autosomal dominant inheritance, congenital heart disease (atrial and ventricular septal defect), radial aplasia or hypoplasia, limb defects frequently asymmetric
VACTERL association	Variable combinations of vertebral, anal, cardiac, tracheal, esophageal, renal, and limb (radial) malformations

Clubhand

In this anomaly, the axis of the wrist is permanently deviated, and the hand is usually closed. Radial clubhand and ulnar clubhand have different prognoses. It is difficult to distinguish between the two in utero; the differential diagnosis depends on the ability to identify the residual forearm bones and the direction of deviation of the wrist (12).

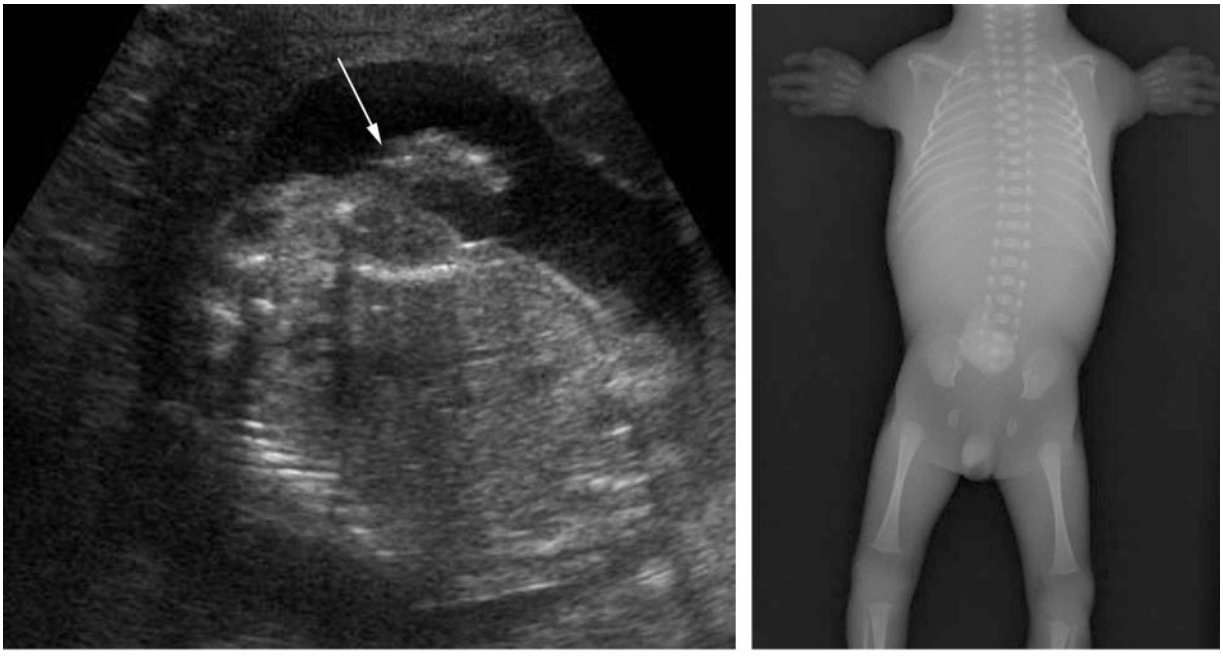
Clubhand with radial deviation of the wrist is the most frequent type detected in utero (Fig 8). Radial clubhand is rarely isolated and sporadic. It usually occurs in association with the radial hypoplasia sequence, which is characterized by preaxial deficits that range from mild hypoplasia of the thumb to complete absence of the radius (12–14). Radial clubhand is frequently syndromic or associated with aneuploidy (Table 2).

Ulnar clubhand is secondary to ulnar deficiency. The diagnosis depends on visualization of the residual radius and the ulnar deviation of the wrist (Fig 9). The anomaly is rare and is usually nonsyndromic but may occur in association with other skeletal anomalies. The differentiation of



a. b.

Figure 9. Ulnar clubhand. **(a)** US image shows fixed deformation of the elbow (arrow) with hand malposition. **(b)** Corresponding radiograph shows a fixed deformation, with the hand clenched and slightly deviated in the ulnar direction because of ulnar aplasia. The deformation of the elbow is secondary to radiohumeral synostosis.



a. b.

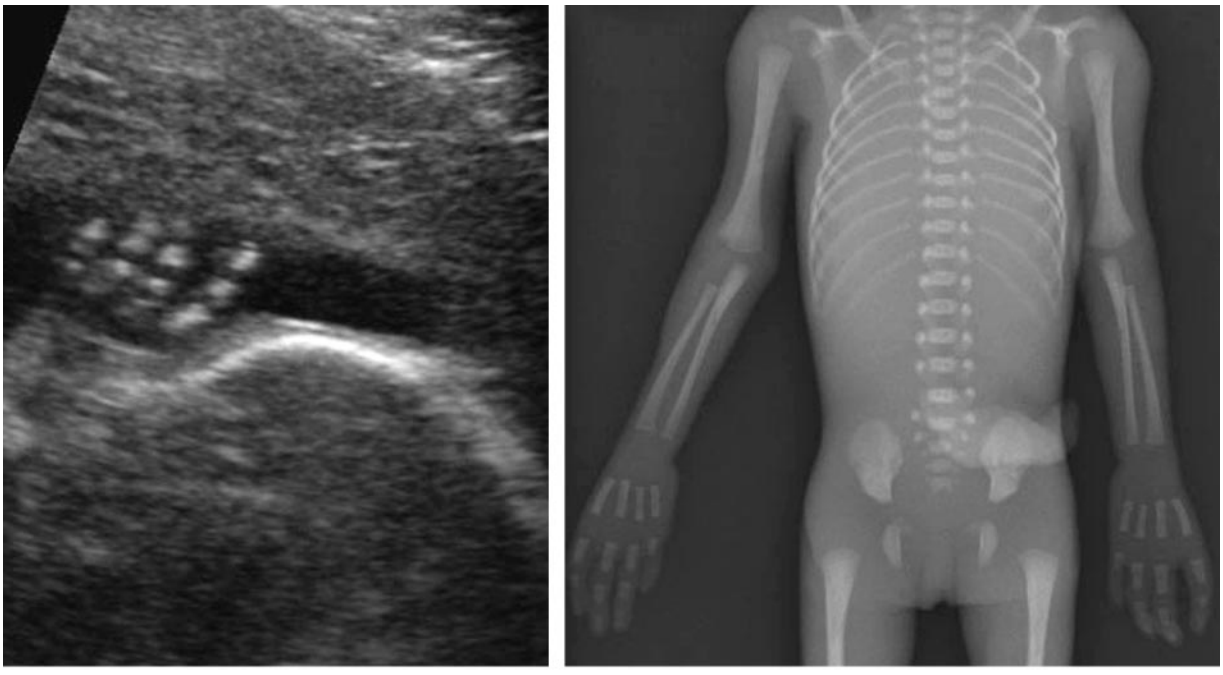
Figure 10. Phocomelia. **(a)** Coronal oblique US image of a fetal thorax at 20 weeks of gestation shows abnormal direct implantation of the hand (arrow) on the thorax. **(b)** Corresponding radiograph shows the bilateral anomaly.

ulnar clubhand from radial clubhand is difficult in utero because the former also may be associated with anomalies of the radial fingers.

Phocomelia

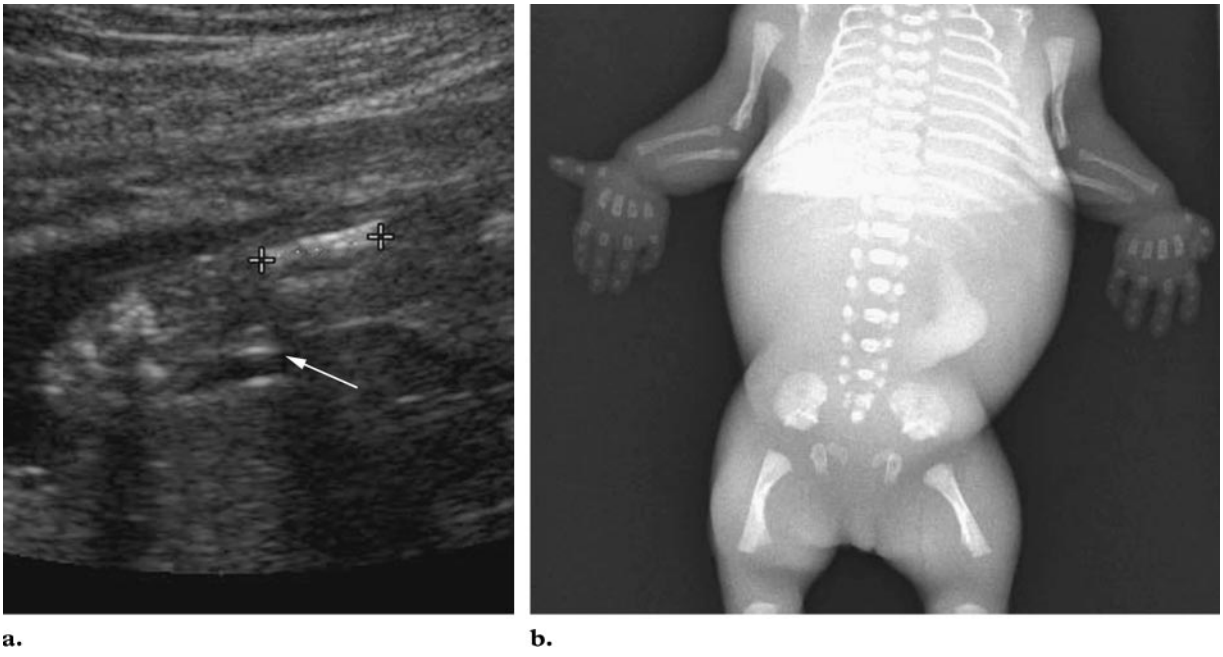
In phocomelia, the hand is present, but the arm or forearm is missing or foreshortened. The hand may be normal or abnormal (Fig 10). Any or all of the limbs may be affected.

Phocomelia can occur sporadically. Historically, thalidomide was a common cause of the anomaly, until use of the drug in women in early pregnancy was banned. (Thalidomide is currently used only in circumscribed settings for its immunomodulatory and antiinflammatory properties.)



a. b.

Figure 11. Thumb aplasia. **(a)** US image of a fetus at 19 weeks of gestation shows only four fingers that are well formed. **(b)** Corresponding radiograph shows the bilateral thumb anomaly.



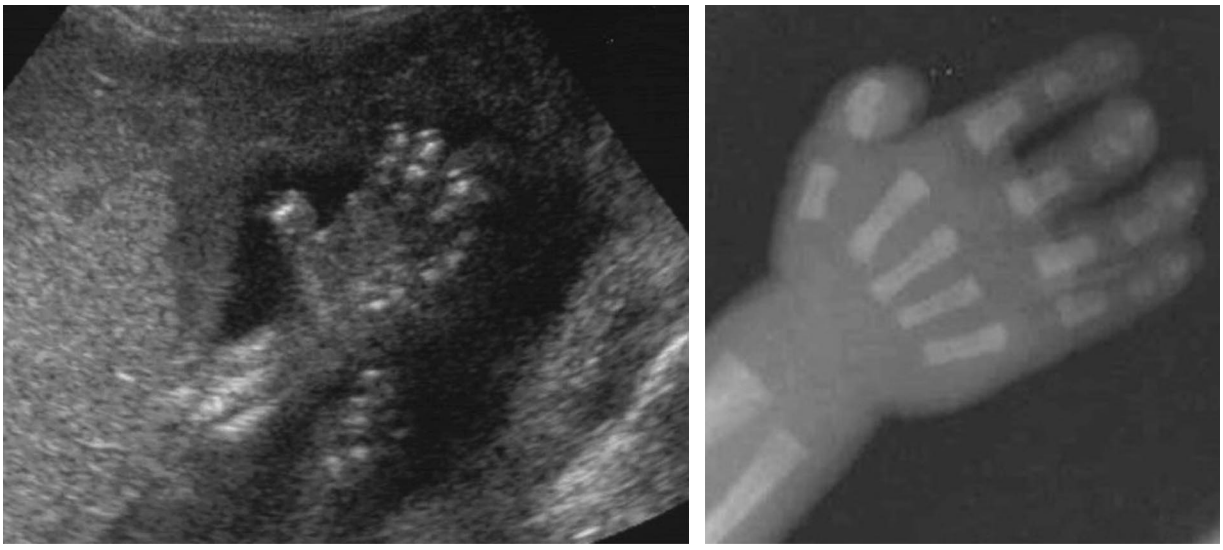
a. b.

Figure 12. Hitchhiker thumb deformation in diastrophic dysplasia. **(a)** US image of a fetus of unknown gestational age shows abnormal abduction of the thumb (arrow) and foreshortening of the long bones (the ulna is visible between the calipers). **(b)** Corresponding radiograph of the same fetus shows the characteristic malposition of the thumbs.

Phocomelia is also associated with Roberts syndrome (an autosomal recessive disorder associated with tetraphocomelia with more severe upper-limb deformities, facial cleft, and polyhydramnios), TAR syndrome, and Grebe syndrome (an autosomal recessive disorder associated with marked hypomelia that is more severe in the lower limbs and that increases in severity distally) (14).

Thumb Anomalies

Thumb anomalies include hypoplasia, triphalangeal thumb, broad thumb, and hitchhiker deformation. These anomalies may be isolated but are more often associated with other hand deformations or syndromes (Figs 11–13) (Table 3).



a. b.

Figure 13. Broad thumb in Pfeiffer syndrome. **(a)** US image at 20 weeks of gestation shows a thickened thumb. This finding, associated with a cranial deformation, allowed a precise prenatal diagnosis of the type of acrocephalosyndactyly. **(b)** Corresponding radiograph helps confirm the hand abnormality.

The extremely rare hitchhiker thumb deformation corresponds to the abnormally abducted position of a more proximally inserted thumb (Fig 12) (14–16). This constant malposition is suggestive of diastrophic dysplasia, a rare bone dysplasia with an autosomal recessive inheritance. The antenatal diagnosis of this anomaly also depends on the identification of micromelic dwarfism of variable severity without thoracic dysplasia and in association with foot deformities (tarsus valgus, metatarsus adductus, clubfoot) and ulnar deviation of the hands (14–16). Flexion contractures lead to multiple subluxations or dislocations. The differential diagnosis includes arthrogryposis and temporary abduction of a normal thumb.

Table 3
Thumb Anomalies

US Finding	Associated Condition or Syndrome
Thumb hypoplasia	Radial hypoplasia sequence
Triphalangeal thumb	Trisomy 13, Fanconi anemia, Holt-Oram syndrome, fetal hydantoin syndrome, Poland sequence, VACTERL association
Hitchhiker thumb	Diastrophic dysplasia
Broad thumb	Acrocephalosyndactylies (Apert, Carpenter, Pfeiffer), trisomy 13

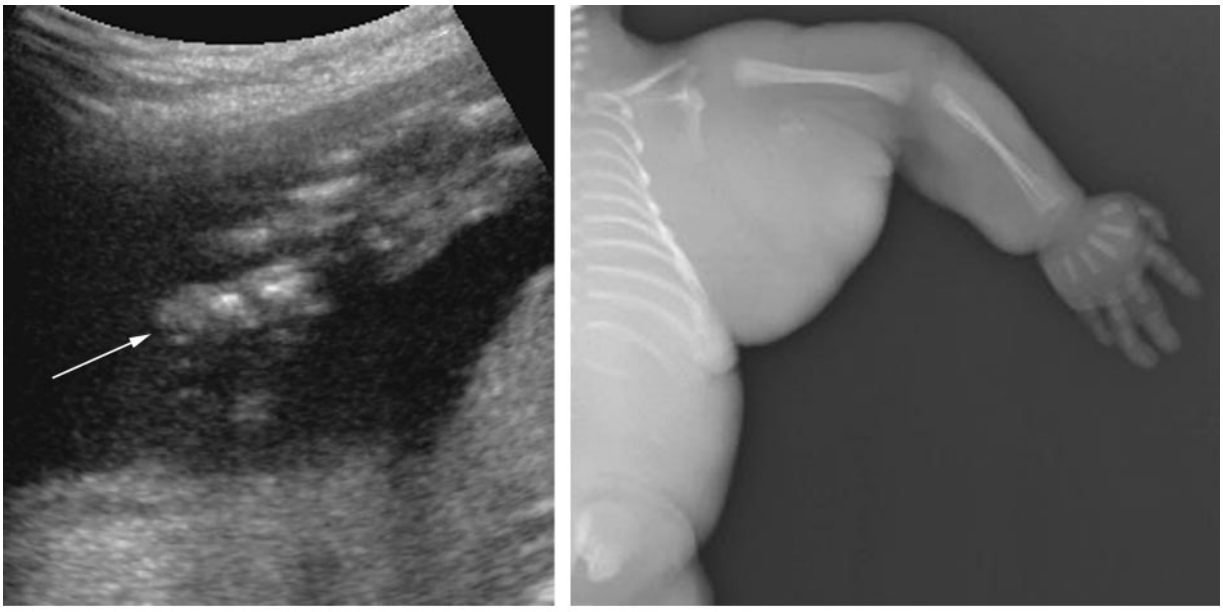
Abnormalities of Size

Macrodactyly

The term *macrodactyly* refers to the overgrowth of all structures in the affected fingers (6). The radial fingers are most often affected. The anomaly is usually isolated but also may occur in association with Proteus syndrome and type 1 neurofibromatosis (Fig 14).

Trident Hand

The last four fingers have the same length in trident hand (5). This anomaly is observed in various chondrodysplasias (Fig 15).



a. b.

Figure 14. Macrodactyly in Proteus syndrome. **(a)** US image at 23 weeks of gestation shows a disproportionately long third left finger (arrow). Massive axillary lymphangioma also was observed at US. **(b)** Corresponding radiograph shows tumefaction of the axillary soft tissues, in association with subtle macrodactyly of the third and fourth fingers.

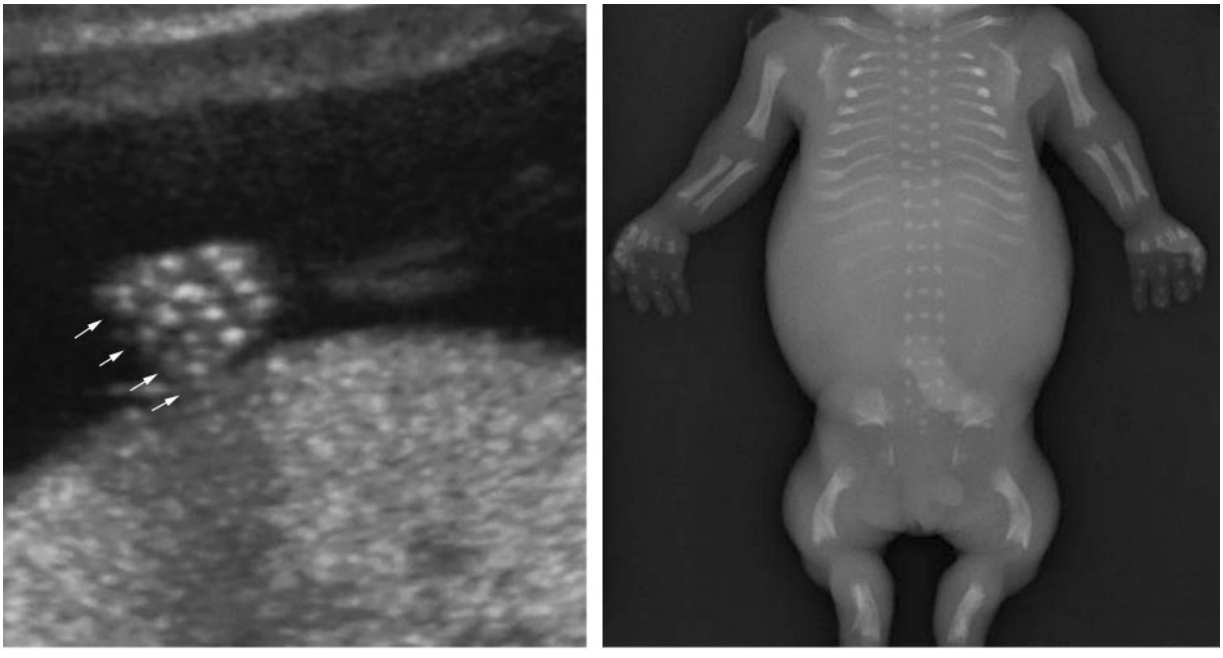


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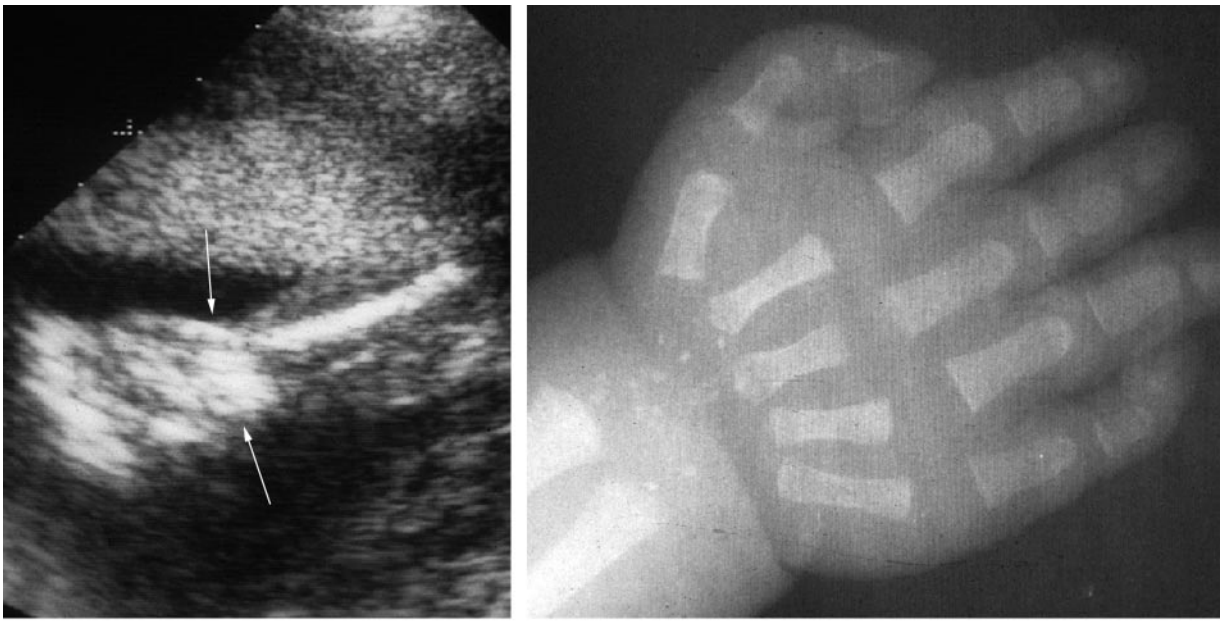
Figure 15. Thanatophoric dysplasia and trident hand. **(a)** US image at 20 weeks of gestation shows four fingers with the same length (arrows). **(b)** Corresponding radiograph helps confirm the hand abnormality.



a. b.

Figure 16. Chondrodysplasia punctata. **(a)** US image of a fetus at 24 weeks of gestation shows abnormal hyperechogenicity of the carpus (arrows). **(b)** Radiograph of the hand of the 1-day-old neonate shows multiple punctate carpal calcifications.

Abnormal Echogenicity Due to Abnormal Calcification

Ossification of the carpus normally occurs after birth. Hyperechogenicity of the carpus at fetal US suggests the presence of abnormal calcification or ossification (Fig 16), which may be due to warfarin exposure, chondrodysplasia punctata, Zellweger syndrome, or alcohol fetopathy (14).

Abnormal Number

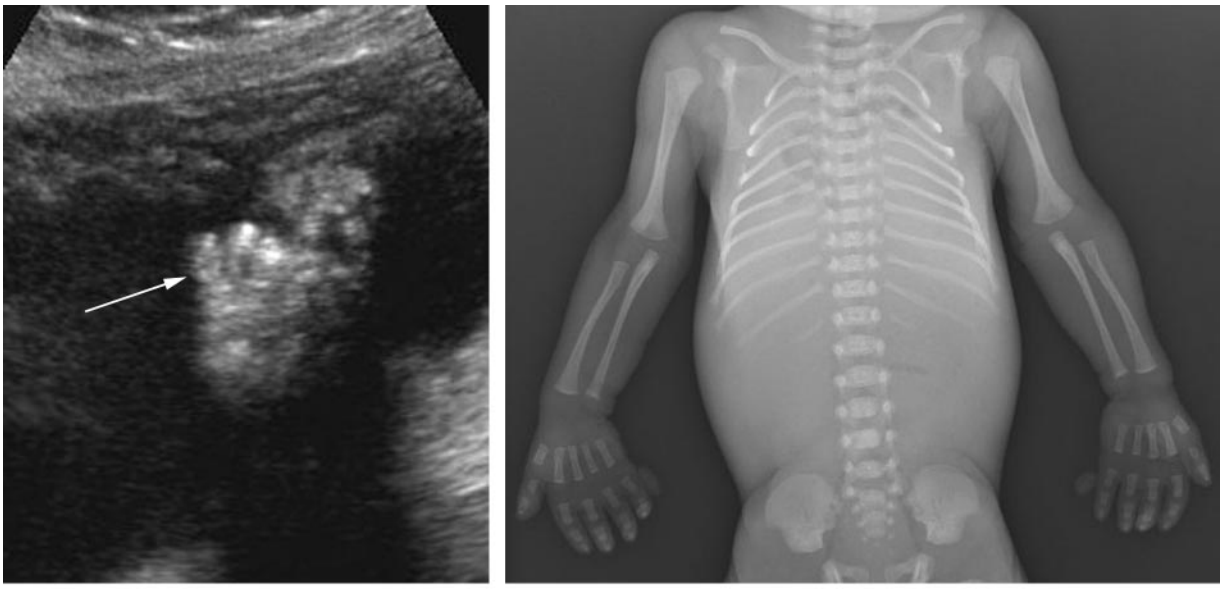
Polydactyly

Polydactyly corresponds to the presence of supernumerary digits that may be more or less well formed. Classification of polydactyly is based on the position of the extra digit on the hand: Polydactyly is described as radial or preaxial when it affects the thumb, ulnar or postaxial when it affects the little finger, and, rarely, central when the three central digits are affected (17). Polysyndactyly corresponds to the association of polydactyly with syndactyly. Polydactyly, the most common hand anomaly, is seen in approximately one of 683 pregnancies (17).

A soft-tissue supernumerary digit is most often an isolated finding and is frequently overlooked at fetal US. In utero amputation of incomplete extra digits has been reported (18). In contrast, a supernumerary finger with an osseous component is more often associated with other anomalies (18).

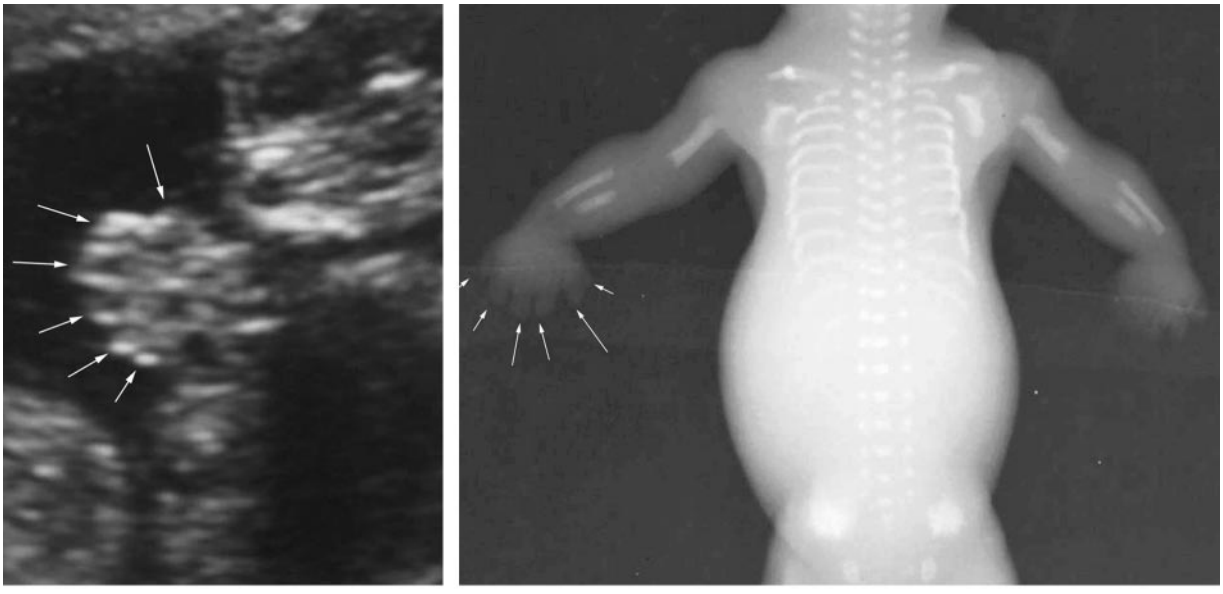
Postaxial polydactyly is more frequent than preaxial polydactyly, particularly among blacks (Fig 17). In most cases, this malformation is an isolated finding with an autosomal dominant transmission, and the prognosis is favorable. However, postaxial polydactyly also may be associated with various syndromes, including trisomy 13 (a fatal condition associated with intrauterine growth retardation and with neural and cardiac malformations), Meckel-Gruber syndrome (also fatal, associated with polycystic kidney disease and posterior encephalocele), Bardet-Biedl syndrome (medullary cystic kidney disease), Smith-Lemli-Opitz syndrome (intrauterine growth retardation and a characteristic high level of 7-dehydrocholesterol), short ribs–polydactyly syndromes (fatal conditions associated with a narrow thorax and short ribs) (Fig 18), and Ellis-van Creveld syndrome (chondroectodermal dysplasia associated with short ribs, mesomelic shortening of long bones, and cardiac defects) (12,14,17–19).

In all these circumstances, the associated anomalies are usually depicted by US. The presence in utero of postaxial polydactyly in associa-



a. b.

Figure 17. Postaxial polydactyly in trisomy 13. **(a)** US image of a fetus of unknown gestational age shows a small incomplete supernumerary digit (arrow) along the fifth finger. **(b)** Corresponding radiograph shows bilateral postaxial polydactyly.



a. b.

Figure 18. Short ribs–polydactyly syndrome. **(a)** US image shows lack of ossification of the hand, with six barely visible digits (arrows). **(b)** Corresponding radiograph, which shows the unossified digits, helps confirm the diagnosis.

tion with renal anomalies but without encephalocele is suggestive of Bardet-Biedl syndrome. Other abnormalities of Bardet-Biedl syndrome, including progressive renal dystrophy, obesity, hypogonadism, and learning difficulties, are usually detected postnatally.

Preaxial polydactyly is less frequently observed. When it is present, a possible syndromic association should be sought, particularly if a triphalangeal thumb is present. Holt-Oram syndrome (atrial and ventricular septal defects,

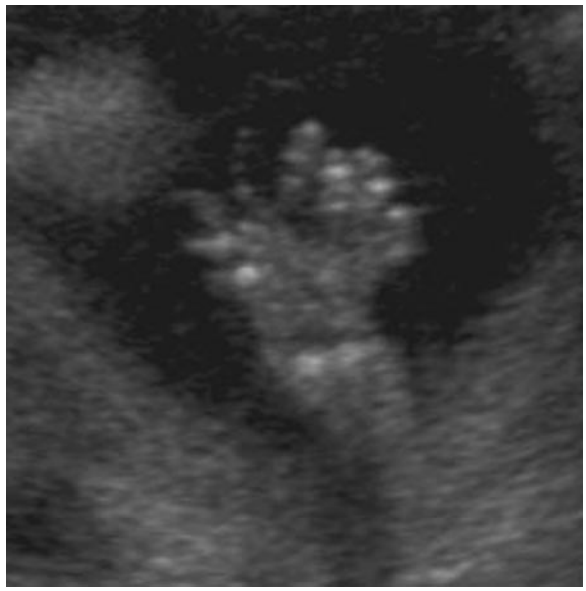


Figure 19. Polydactyly of the central type. US image of a fetus at 20 weeks of gestation shows six clearly identifiable fingers.

Table 4
Syndromic Fetal Craniostenosis

Syndrome	US Characteristics
Apert syndrome	Acrocephalosyndactyly (Apert type), acrocephaly, severe complex syndactyly of the last four digits or all the fingers and toes (cupped, mittenlike hands)
Carpenter syndrome	Acrocephalosyndactyly (Carpenter type), brachymesophalangia, soft-tissue syndactyly (hands and feet), preaxial polydactyly of the feet
Pfeiffer syndrome	Acrocephalosyndactyly (Pfeiffer type), broad thumbs and toes, mild soft-tissue syndactyly
Crouzon syndrome	Craniofacial dysostosis, no limb anomaly

abnormal thumb [from absent to triphalangeal]), short ribs–polydactyly syndromes, Carpenter syndrome, trisomy 21, VACTERL association, and Fanconi anemia are conditions that may be associated with preaxial polydactyly.

Central polydactyly, the least frequent type of polydactyly, is often associated with another malformation or a syndrome (Fig 19).

Syndactyly

Syndactyly, an abnormal connection between adjacent digits, is described according to its extent (6). The fusion can involve only soft tissues (simple) or include bones (complex). It may be either complete, along the entire length of the finger, or incomplete, with sparing of the distal part of the fingers. Any number of digits can be linked. The second and third digits are the most frequently affected.

Simple syndactyly is difficult to diagnose in utero: The affected fingers move together and are never seen independently (Fig 20). Complex syndactyly is more obvious because the affected fingers and phalanges are deformed. When syndactyly affects more than two fingers, as in Apert syndrome, the hand has a strange appearance (spade-, spoon-, or mitten-shaped), and the fingers cannot be clearly individualized (Fig 21).

Syndactyly is relatively common; it affects two to three of 10,000 live births. The anomaly can be sporadic, familial, or associated with other abnormalities. Familial syndactyly usually affects the second and third digits and is not associated with other abnormalities; transmission is autosomal dominant, with variable expressivity and incomplete penetrance. Associated or complicated syndactyly can be syndromic or secondary to a constriction band sequence (14). Syndromes described in association with complicated syndactyly include acrocephalosyndactylies and the Poland sequence.

In the presence of syndromic craniostenosis, a detailed assessment of the hands is essential to reach the correct diagnosis (Table 4).

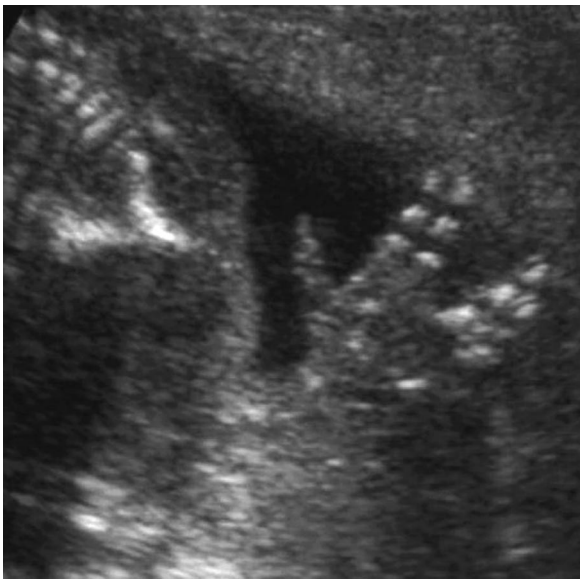
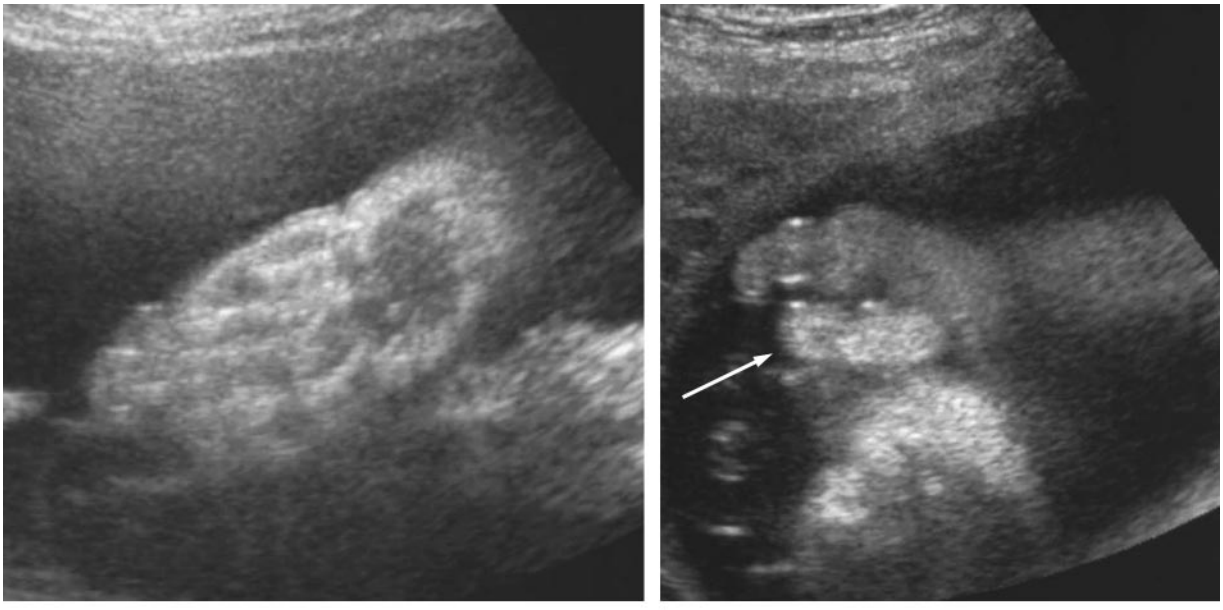
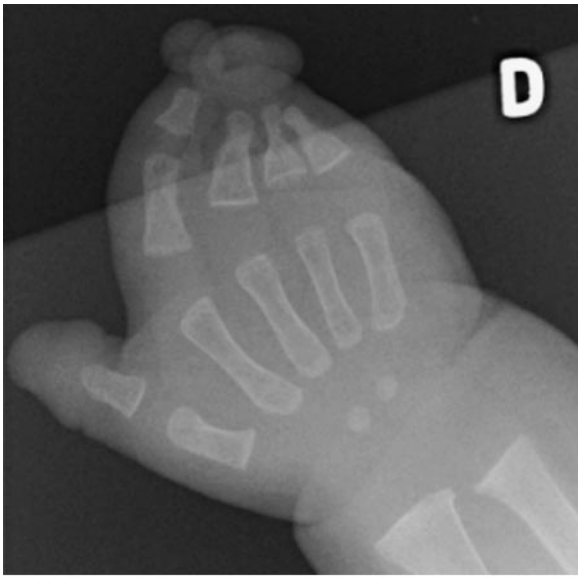


Figure 20. Soft-tissue syndactyly. US image of a fetus at 18 weeks of gestation shows soft-tissue syndactyly of the fourth and fifth fingers, which remained linked throughout the US examination, while the other fingers moved independently.



a. **b.**



c.

Figure 21. Complex syndactyly. **(a)** US image of a fetus at 34 weeks of gestation shows the last four digits stuck together, with a mittenlike appearance of the hand. **(b)** US image shows that the thumb (arrow) is independent; the fetus is able to grip. **(c)** Corresponding radiograph of the hand at 4 months of age.

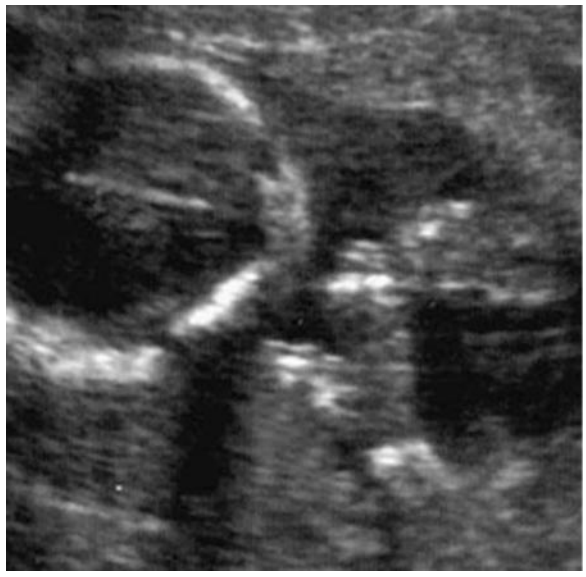
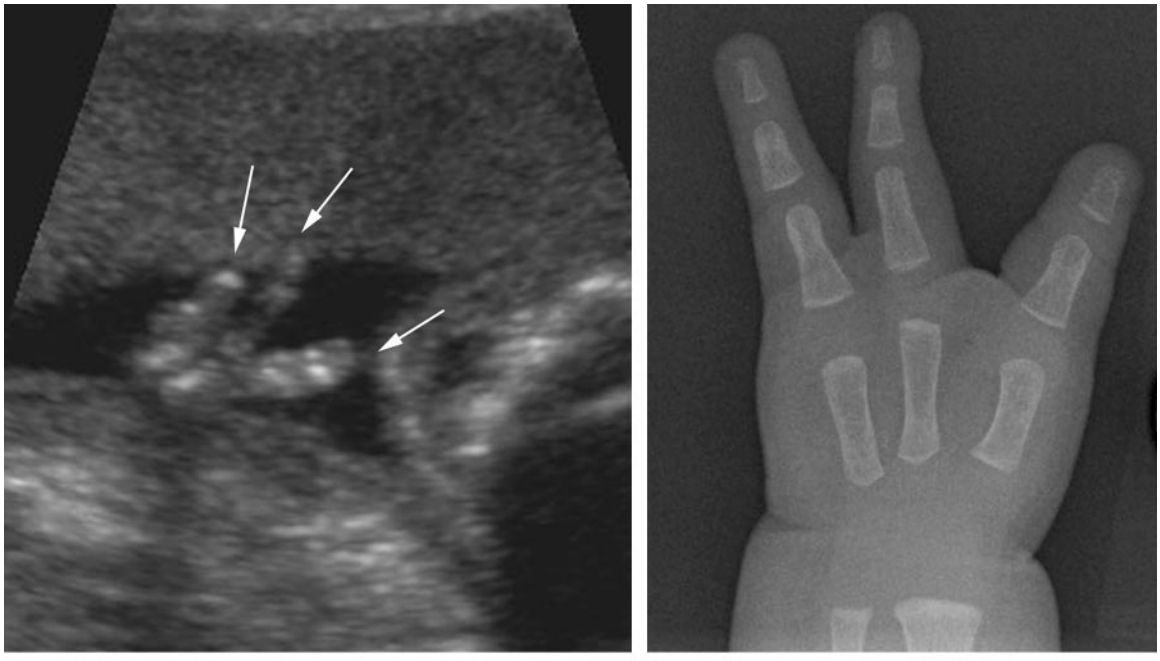


Figure 22. Ectrodactyly in split-hand and -foot syndrome. US image at 20 weeks of gestation shows a deep central V-shaped cleft in the hand.



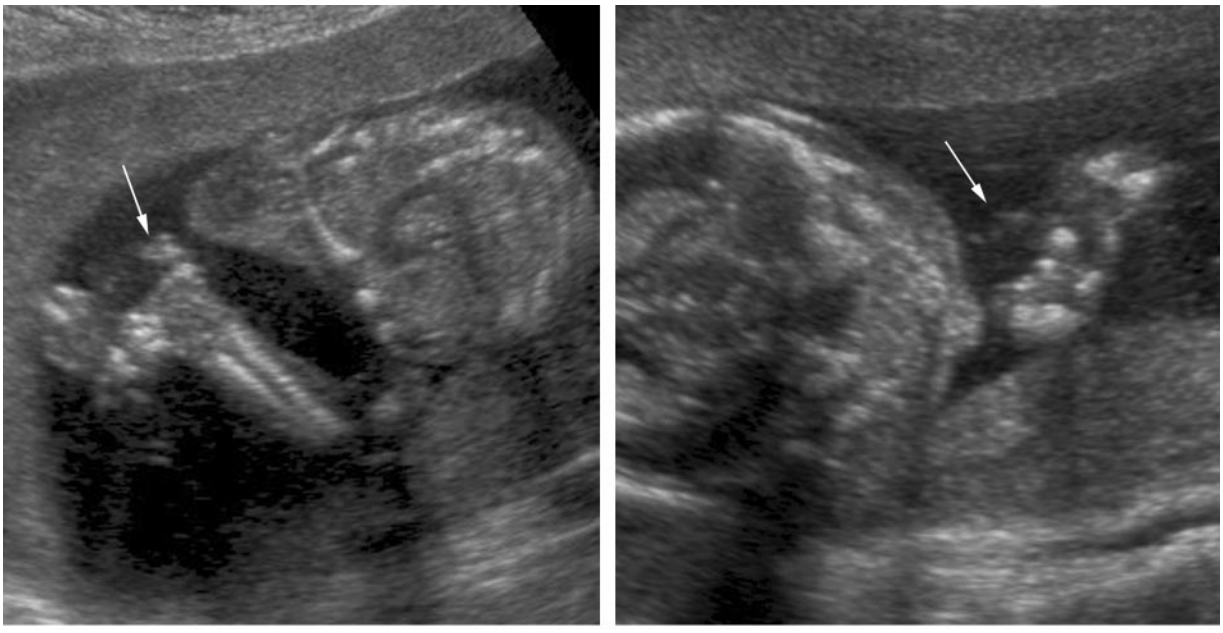
a. **b.**

Figure 24. Oligodactyly. **(a)** US image at 21 weeks of gestation shows only three fingers (arrows). **(b)** Corresponding radiograph after birth shows the absence of the fourth and fifth fingers.

Ectrodactyly (Split Hand)

Split or cleft hand (also known as lobster claw hand) results from a longitudinal deficiency of the central digits. The main pathogenic mechanism is probably a failure of the median apical ectodermal ridge in the developing limb bud (20).

The malformation is characterized by a deep V- or U-shaped central defect. It may be associated with syndactyly, aplasia, or hypoplasia of the residual phalanges and metacarpals. Hands and feet can be affected (split-hand and -foot malformation) (Fig 22). The severity of the malformation is highly variable (Fig 23). Split hand



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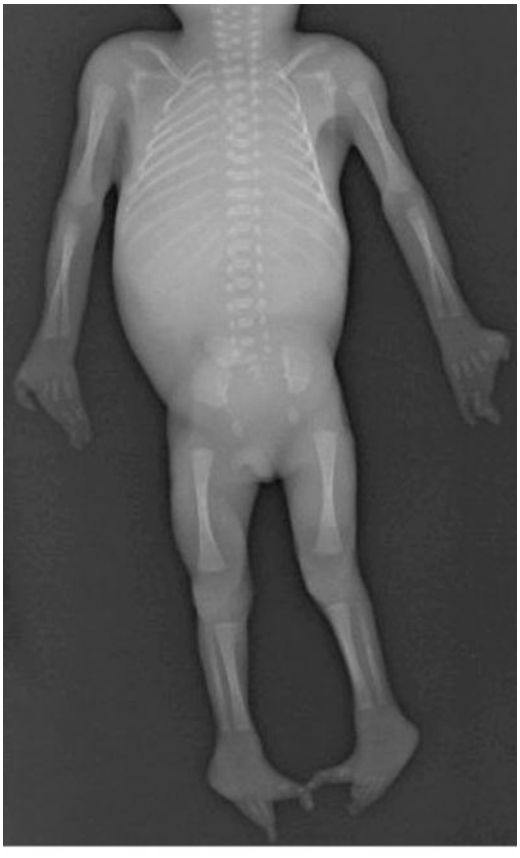


c.

Figure 23. Ectrodactyly. **(a)** US image of a fetus at 19 weeks of gestation shows a wide gap between the thumb (arrow) and the last two fingers of the left hand, which are malformed. **(b)** US image of the same fetus shows the right hand, which also is affected by wide ectrodactyly (the arrow indicates the thumb) in association with syndactyly of the last two digits. The fetus also manifested cleft lip, cleft palate, and micrognathia. **(c)** Corresponding fetal radiograph shows ectrodactyly of the four extremities.

may be isolated or associated with syndromes such as EEC (*e*ctrodactyly, *e*ctodermal dysplasia, and *c*left lip or palate), Roberts syndrome, and others (20). The most common mode of transmission is autosomal dominant, but autosomal recessive and X-linked transmission also have been described.

The differential diagnosis of split hand includes oligodactyly (a reduced number of well-formed fingers) (Fig 24) and constrictive amniotic bands.

Congenital Constriction Band Sequence

The estimated incidence of the amniotic band sequence varies from one of 56 fetuses to one of 15,000 live births, to one of 1200 live births. The cause of this abnormality is still unknown, but two possibilities have been proposed: vascular disruption events and early amnion rupture leading to constrictive amniotic bands (21–23).

This entity covers a wide spectrum of anomalies, from simple grooves to ring constriction, amputation, syndactyly, pterygium, facial cleft, and cranial defect. The anomalies are characteristically asymmetric (11).

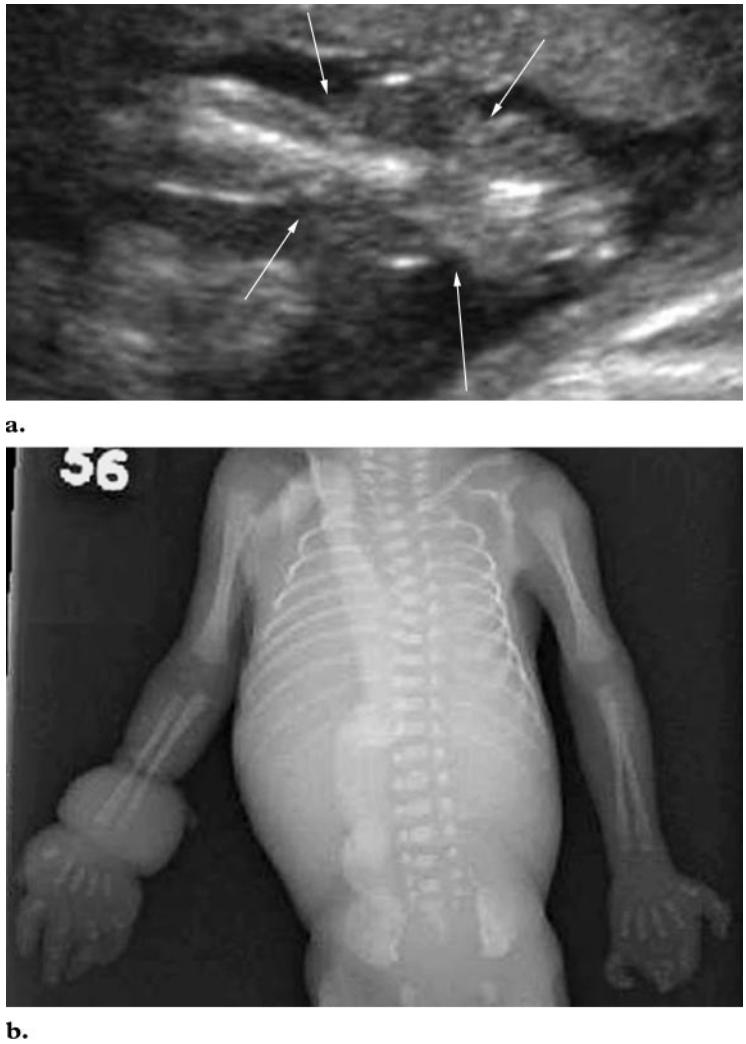
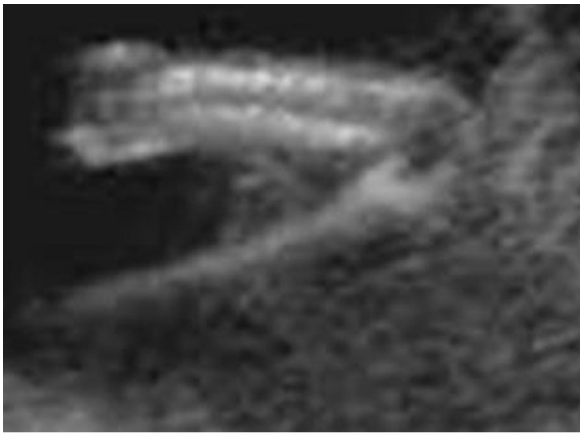


Figure 25. Congenital constriction band sequence. **(a)** US image of a fetus at 18 weeks of gestation shows constrictive rings (arrows) of the right forearm, with distal soft-tissue edema. **(b)** Corresponding fetal radiograph helps confirm the presence of a constrictive ring of the right forearm and shows distal amputation and syndactyly on the left side.

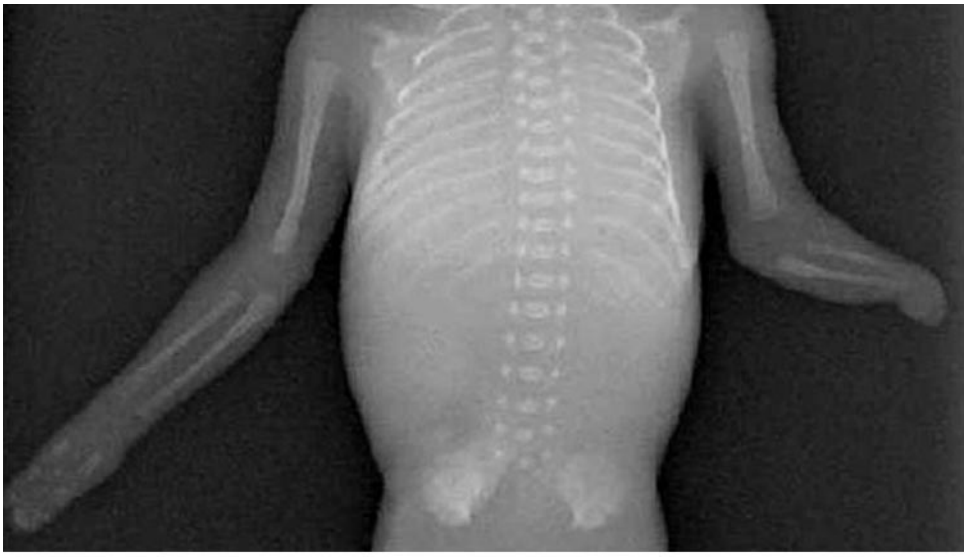
Prenatal diagnosis depends on the discovery of the malformation, which is rarely associated with loose adhesive amniotic bands (Figs 25, 26). Ring constriction associated with edema of the distal part of the limb can lead to progressive amputation in utero. Congenital constriction band sequence is usually sporadic (11).

Distal amputation or hypoplasia of the central digits (especially the third) can also be secondary to early chorionic villus sampling. Vascular disruption has been postulated as the cause (24).

The differential diagnosis of congenital constriction band sequence includes the Adams-Oliver syndrome and other congenital transverse deficiencies unrelated to band constriction. In Adams-Oliver syndrome (aplasia cutis, limb defects), scalp and skull defects at the vertex are associated with transverse limb defects, which are often asymmetric. This condition, which has an autosomal dominant mode of inheritance, is usually diagnosed after birth.



a.



b.

Figure 26. Congenital constriction band sequence. **(a)** US image of a fetus at 19 weeks of gestation shows distal amputation at the level of the left wrist. **(b)** Corresponding fetal radiograph confirms amputation.

Conclusions

Assessment of the fetal hands should be part of a complete fetal US examination. Fetal hand malformations have major implications for functional prognosis. They can be isolated but are more often indicative of syndromic associations or karyotype anomalies. Most malformations are classifiable according to well-established groups, which helps to narrow the differential diagnosis. Sometimes, however, the complexity of the deformations defies their categorization. A multidisciplinary approach is thus mandatory to review and integrate the imaging findings and the results of complementary examinations.

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Obstetric US: Watch the Fetal Hands

Françoise Rypens, MD, et al

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Page 812
Hand anomalies encompass a large spectrum of malformations, from subtle defects of distal phalanges, almost undetectable in utero, to severe abnormalities such as clubhand, split hand, or absence of a limb.

Page 812
A systematic examination of the fetal extremities is worth the effort, because knowledge of fetal hand anomalies can help guide the diagnosis and management of many associated conditions (3).

Page 812
The discovery of a hand malformation can help to narrow the differential diagnosis. Some hand malformations are even specific.

Page 812
If a fetal hand anomaly is detected, the patient should be referred to a clinic that specializes in the identification and management of fetal malformations and genetic syndromes. A complete fetal work-up is necessary.

Page 813
Fixed positions of the wrists or the digits are abnormal. When an alignment abnormality is fixed, the prognosis is often guarded, not only because of the functional effects but also because of the frequent association of such fixed positions with karyotype anomalies and syndromes (Table 1).

Table 1
Alignment Abnormalities

US Finding	Associated Condition or Syndrome
Clenched hand	Trisomy 18, FADS
Camptodactyly	Trisomies (18, 13, 15), FADS
Clinodactyly	Trisomy 21
Clubhand	Aneuploidies, Fanconi anemia, TAR syndrome, Aase syndrome, Holt-Oram syndrome, VACTERL association
Phocomelia	Sporadic, thalidomide use, Roberts syndrome, TAR syndrome, Grebe syndrome

Note.—FADS = fetal akinesia deformation sequence, VACTERL = *v*ertebral, *a*nal, *c*ardiac, *t*racheal, *e*sophageal, *r*enal, and *l*imb abnormalities.