Can you give me a hand? Diagnosing and understanding the clinical significance of fetal hand anomalies in obstetric ultrasound

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Background and Introduction

- Fetal hand anomalies encompass a large spectrum of malformations which are commonly missed during routine fetal ultrasonography.

- Hand anomalies can help guide the diagnosis and management of many associated conditions.

- If a fetal hand anomaly is detected, consider referral to a clinic that specializes in the identification and management of fetal malformations and genetic syndromes.
  - A detailed fetal ultrasound (US) with careful cardiac evaluation to determine the presence or absence of associated abnormalities.
  - Repeated focused US examinations are sometimes necessary to diagnose more precisely the underlying anomalies as pregnancy progresses.

- Various systems exist for the classification of upper limb malformations on the basis of anatomy, embryology, genetics, and teratology.
Materials and Methods

- Retrospective review of fetal hand anomaly cases at OHSU from 2008-2015.
- 8 fetal hand anomalies are presented and associations discussed, with post delivery follow-up.

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Polydactyly

Terminology:
• Postaxial: Ulnar or fibular side.
• Preaxial: Radial or tibial side (less common).

General features:
• Spectrum of appearances based on formation of digit
  - Complete → Bifid → Broad digit
  - Soft tissue "nubbin" (digiti postminimi)
  - Triphalangeal thumb

Ultrasound Findings:
• Confirm in both axial and coronal views +/- 3D
• Extra digit may be hypoplastic or angulated in position
• If soft tissue without bone, often missed prenataally

Associations:
• Familial (isolated), Trisomy 13 and 18, Meckel-Gruber syndrome, Diabetic Embryopathy

3D US shows postaxial polydactyly with a 6th digit on the ulnar side of the hand (arrow). A midline cleft lip is noted (curved arrow) in this fetus with trisomy 13. T=thumb

Prenatal 3D US and postnatal radiograph demonstrate preaxial polydactyly adjacent to the thumb (arrow).

Postnatal radiograph shows a postaxial digiti postminimi, not detected in utero (arrow).
Terminology:
• Radial deviation of the distal 5th digit.
• Short middle phalanx

Imaging:
• Seen best on coronal open hand view in 2nd trimester.

Ultrasound Findings:
• Tip of 5th finger curves toward 4th finger

Associations:
• Familial clinodactyly
  • Autosomal dominant
• Minor marker for trisomy 21
  • 60% of T21 have clinodactyly

Gray scale ultrasound, 3D ultrasound, and photographic images of the right hand demonstrate radial deviation of the distal 5th digit.
Clenched hand

Terminology:
• Fetal hands are held in a clenched position as if unable to extend.

Pathology:
• Muscle variations along the radial margin of the forearm and hand.
• Absence of thenar muscles with anomalous tendons and attachments among the forearm muscle groups

Ultrasound Findings:
• Hands are in persistent clenched position.
• May have overlapping fetal fingers (particularly index finger overlapping the middle finger).

Associations:
• Present in 50% of Trisomy 18 cases
• Can be seen in fetal akinesia deformation sequence (arthrogryposis)
Syndactyly

Terminology:
- Partial or incomplete syndactyly: Affects only proximal segment of digit.
- Complete syndactyly: Affects entire length of digit to nail.

Pathology:
- Failure of separation of digital rays.
- Occurs in the first trimester

Imaging:
- Seen best on coronal open hand view in 2nd trimester.
- 3D imaging often helpful to further evaluate digits.

Ultrasound Findings:
- Inability to see separated digits on open hand view of fetus.
- Often missed on prenatal US due to variable fetal hand position

Associations: Non-syndromal (Familial), Amniotic Band Syndrome, Apert Syndrome, Trisomy, Poland Syndrome

Prenatal gray-scale US and postnatal AP radiograph show complete syndactyly of the 3rd-5th digits (arrows).
Arthrogryposis

Terminology:
• Abnormality related to lack of fetal movement.

Ultrasound findings:
• Lack of fetal motion
• Polyhydramnios: Decreased fetal swallowing.
• Abnormal posturing:
  • Cross-legged lower limbs
  • Extended elbows with internally rotated, flexed wrists (“waiters tip”).
  • Clubfeet
  • Clenched hands.

Associations:
• Trisomy 18
• Distal Arthrogryposis
• Amyoplasia
• Multiple pterygium syndrome

3D ultrasound shows a fetus with flexed wrists (arrow) and hyperextended fingers (curved arrow).

Clinical photograph of the newborn shows flexed wrist and thin curved fingers.

3D ultrasound shows a fetus with extended elbows (arrow) and rotated flexed wrists (curved arrow).
Radial Ray Syndrome

Terminology:
• Spectrum of anomalies including absence or hypoplasia of radius, radial carpal bones, or thumb.

Imaging:
• Detectable on routine anatomic survey or earlier.

Ultrasound Findings:
• Radius is absent or hypoplastic
• Radial deviation of the hand
• Thumb may be absent or hypoplastic
• Proximal implantation of the thumb with associated triphalangeal appearance.

Associations
• VACTERL, Trisomy 13 and 18, Holt Oram Syndrome

3D and gray-scale images show short left radius (open arrow), thumb and second digit. Three digits are present with abnormal angulation at the wrist (arrow).
Ectrodaclyly

Terminology:
• Characterized by hypoplasia of phalanges, metacarpals, metatarsals, and deep median cleft with fusion of remaining digits.
• Type of split hand/foot malformation.

Ultrasound findings:
• Cleft appearance of hands and/or feet with missing digits.
• Variable clefting from just soft tissue to deep cleft and medial ray deficiency.
• Syndactyly of digits on either side of cleft often seen (soft tissue and/or osseous fusion).

Associations:
• Ectrodactyly-Ectodermal Dysplasia Clefting Syndrome (ECC)

3D fetal ultrasound and AP radiograph show a cleft of the hand secondary to a missing middle metacarpal and phalange (arrow).

Clinical photograph of the left hand shows central ray deficiency and deep cleft (arrow).
Amniotic Band Syndrome

Terminology:
• Entrapment of fetal parts by disrupted amnion.
• Formation of fibrous amniotic bands which can result in multiple anomalies (clefts, constrictions, amputations, malformation, deformation)

Imaging:
• Asymmetric distribution of defects is hallmark
• Edema of distal extremity secondary to constriction.

Ultrasound Findings:
• Head/face clefts or defects
• Truncal entrapment
• Extremity entrapment

Associations:
• Sporatic occurrence most common
• Ehlers-Danlos syndrome
• Epidermolysis bullosa

3D fetal ultrasound and postnatal clinical photo show amputation of the 2nd-4th digits (arrow) with intact thumb (T).
Conclusion

- Identifying and characterizing fetal hand anomalies can be challenging, but is important to attempt at the time of the routine anatomy scan.

- When found prenatally, careful evaluation for associated anatomic abnormalities or dysmorphic conditions should be performed.

- A significant portion of fetuses with hand anomalies will have additional findings.
References


