Congenital upper limb malformations: pictorial review

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Authors: J. Aucourt¹, J.-F. BUDZIK², S. Manouvrier³, A. Mezel³, M. Cagneaux⁴, N. Boutry³, ¹lille/FR, ²La Madeleine/FR, ³Lille/FR, ⁴Marcq En Baroeul/FR  
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Learning objectives

To illustrate common and rare congenital abnormalities of the upper limb and to review their terminology and classification.

To present the main embryofoetopathies, genetic disorders and non genetic syndromes encountered in this pathology.

Background

Upper limbs malformations occur in approximatively 6.5 on 10000 neonates. They are complex and knowledge of their radiological and clinical features is essential both for diagnosis and treatment planning.

Congenital upper limb malformations are divided into 1) failure of formation with transverse, intercalary and longitudinal deficiencies (preaxial, centroaxial, postaxial), 2) failure of differenciation (synostosis, syndactyly), 3) duplication (polydactyly) and 4) brachydactyly. Sprengel deformity and congenital pseudarthrosis of the clavicle are also described.

This pictorial review is also an opportunity to cover the topics of clinodactyly, camptodactyly and Madelung's deformity that are usually included in congenital upper limb malformations.

Other skeletal and non skeletal abnormalities must be considered to eventually diagnose:

- Embryofoetopathy (maternal valproate treatment, constriction band syndrome),
- Genetic disorders (trisomy 21, Turner syndrome, Holt Oram syndrome),
- And non genetic syndromes (VACTERL, Poland).

Imaging findings OR Procedure details

I- Failure of formation

Transverse deficiency (fig 1):
Transverse deficiency may be divided into amelia (absence of arm), hemimelia (absence of forearm), acheiria (absence of hand) and symbrachydactyly. They are commonly sporadic, due to a vascular accident during morphogenesis. They may occur in syndromes such as tetraamelie, Roberts syndrome (facial abnormalities), Hanhart syndrome (hypoglossia-hypodactylyia) and toxic embryofoetopathies.

Amniotic band syndrome is a differential diagnosis of transverse deficiency. It represents a disruption or deformation sequence subsequent to annular bands of chorionic tissue encircling the limb. It has a low incidence of associated abnormalities, normal proximal anatomy, and a sporadic occurrence. Clubfoot is the most common associated abnormality. Distal fusion of amputated digits, as in acrosyndactyly, may be present. Radiological findings are characteristic and show amputation below an annular band.

Preaxial deficiency (figs 2 and 3)

It may vary from minor abnormalities of the thenar muscles to complete absence of the preaxial structures (radius, radial carpal bones, and thumb). The most common presentation is a short radius, absence of scaphoid and trapezium, and total absence or hypoplasia of the thumb (ie, radial club hand). Coalition of radial carpal bones, proximal radioulnar synostosis and radial head dislocation may be seen. Preaxial deficiency may occur bilaterally or unilaterally and is often part of a syndrom.

VACTERL (Vertebral abnormalities, Anorectal atresia, Cardiac defects, Tracheo-oesophageal fistula with or without Esophageal atresia, Renal abnormalities, Limb abnormalities)

Fanconi's anemia (aplastic anemia not present at birth)

TAR syndrome (thrombocytopenia- absence of radius)

Holt-Oram syndrome (heart defects, most commonly septal defects)

Nager syndrome (craniofacial malformations)

Baller Géroid syndrome (craniosynostosis)

Okihiro syndrome (eye abnormalities)

Trisomy 13 or 18

Main syndromes associated with preaxial deficiency

Postaxial deficiency (fig 4)
It is characterized by ulnar or postaxial digit (IV/V) deficiencies. Humeroradial synostosis, radial head dislocation, carpal or metacarpal (IV-V) coalition and pre-axial digit (I/II) deficiencies may be seen. Most of them show autosomal dominant (AD) patterns of inheritance. Syndromic associations are rare. Postaxial deficiencies usually are unilateral compared to preaxial deficiencies.

**Miller syndrome** (craniofacial malformations)

**Cornelia De Lange syndrome** (distinctive facial features and multiple congenital malformations)

**FFU syndrome** (Femur Fibula Ulna syndrome)

**Main syndromes associated with postaxial deficiency**

Centroaxial deficiency (fig 5)

It manifests as absence of digits or metacarpals within the central portion of the hand. Carpal coalition, pre- or postaxial syndactyly, surnumerary metacarpal bone may be seen. The radius and ulna are normal. Centroaxial deficiency may be classified as being either typical or atypical. The typical cleft hand is generally bilateral and familial whereas the atypical one is unilateral and spontaneous.

**SHFM syndrome** (Split Hand Foot Malformation)

**EEC syndrome** (Ectrodactyly, Ectodermal dysplasia, Clesting syndrome)

**EEM syndrome** (Ectrodactyly, Ectodermal dysplasia, Macular dystrophy)

**Main syndromes associated with typical central deficiency**

Intercalary deficiency (fig 6)

In phocomelia, a functional terminal element is always present. Different types of phocomelia are as follows: those in which the hand attaches to the shoulder (forearm and arm deficient), those in which the forearm attaches to the shoulder (arm deficient) and those in which the hand attaches to the arm (forearm deficient). The arm deficient type is often spontaneous whereas the two others may occur in toxic embryofetopathy or Roberts syndrome.

**II- Failure of differenciation**

Humeroradial synostosis (fig 4)
It is often encountered in postaxial deficiency.

Radioulnar synostosis (fig 7)

It is often bilateral and spontaneous. It may also occur in preaxial deficiency and Klinefelter syndrome.

Syndactyly (fig 8)

Syndactyly is classified according to completeness (complete, incomplete) and presence of bony union (simple, complex, complicated). Complicated syndactyly refers to any syndactyly in which more than a simple side-side bony fusion exists. They may be classified in preaxial, postaxial, centroaxial, complete and synpolydactyly.

<table>
<thead>
<tr>
<th>Syndactyly</th>
<th>Main causes</th>
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<tbody>
<tr>
<td>Preaxial (I-II)</td>
<td>Preaxial syndromes (Holt Oram, Townes Brocks syndromes...)</td>
</tr>
<tr>
<td>Centroaxial (III-IV), the most common form</td>
<td>Spontaneous</td>
</tr>
<tr>
<td>Postaxial (IV-V)</td>
<td>Familial (dominant) or ODD syndrome</td>
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<tr>
<td>Complete</td>
<td>Apert syndrome (fig 9)(craniostenosis) and other acrocephalosyndactylies</td>
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<tr>
<td>Synpolydactyly (mesoaxial syndactyly with duplication of the fourth digit)</td>
<td>Familial (HOXD13 gene)</td>
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**Classification and causes of syndactylies**

Apert syndrome is an autosomal dominant disorder. It is the most severe form of acrocephalosyndactyly. The main radiographic sign is a symetrical complete syndactylie of hands and feet (mitten hands and sock feet). It is associated with craniosynostosis.

**III- Duplication**

Preaxial polydactyly (duplicated thumb) is the most common form and the level of duplication is variable. It is commonly spontaneous but syndromes may be associated (fig 10).

Postaxial polydactyly is characterized by fifth digit duplication in hands and/or feet. Two phenotypic varieties have been described. In type A, the extra digit is well formed and articulates with the fifth or an extra metacarpal. A rudimentary extra fifth digit characterizes the type B. Postaxial polydactyly has a strong AD inheritance pattern and
is commonly seen in individuals of African descent. It may also be seen in syndromes (fig 11).

Central polydactyly is rare. Complex duplication is also described. Polysyndactyly is an example (fig 12).

<table>
<thead>
<tr>
<th>Preaxial polydactyly</th>
<th>Preaxial syndromes</th>
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<tbody>
<tr>
<td></td>
<td>Rubinstein-Taybi syndrome</td>
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<td></td>
<td>Acrocephalosyndactyly</td>
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<tr>
<th>Postaxial polydactyly</th>
<th>Smith-Lemli-Opitz syndrome</th>
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<td>Bardet-Biedl syndrome</td>
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<td>Ellis Van Creveld syndrome</td>
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<td>Pallister-Hall syndrome</td>
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</table>

<table>
<thead>
<tr>
<th>Centroaxial polydactyly</th>
<th>Pallister-Hall syndrome</th>
</tr>
</thead>
</table>

| Complex polydactyly | Greig syndrome |

**Main syndromes associated with polydactyly**

**IV- Brachydactyly**

It is divided into 5 types (fig 13,14). Brachymesophalangy may be responsible for clinodactyly. Brachydactyly A3 is considered as an anatomic variant. Brachydactyly C (brachymesophalangy of the index, middle and little fingers with shortening of the first metacarpal) may be very heterogeneous: hyperphalangy of the index, anomalous configuration of the index (resulting in its ulnar deflection), short metacarpals, symphalangism and "angel-shaped" phalanges (fig 15) are occasionally present.

Brachydactylies are commonly isolated malformations but they may be part of a complex malformation. The exception is brachydactyly E in which Albright osteodystrophy and Turner syndrome are potential diagnoses (fig 16).

**V- Sprengel deformity** (fig 17)

It is a failure of the scapula descent from the level of the embryonic limb bud opposite the fifth cervical vertebra to its thoracic position. Hypoplasia of the scapula
and associated abnormalities are commonly seen (of the clavicles, ribs, vertebrae and shoulder musculature). The most common associated finding is the omovertebral bone.

**VI- Congenital pseudarthrosis of the clavicle** (fig 18)

Congenital pseudarthrosis of the clavicle usually occurs on the right side. Radiologic evaluation shows lack of bone continuity in the middle third of the clavicle without evidence of reactive bone. It needs to be distinguished from birth fracture, neurofibromatosis and cleidocraniodysostosis.

**VII- Clinodactyly** (fig 19)

It denotes a deviation of a finger as a result of an abnormally shaped middle phalanx. It may occur because of a delta phalanx. The latter corresponds to a congenital triangular bone. The classic finding is seen as a bone with a continuous C-shaped physis extending along the length of the shortened side, like a staple.

**VIII- Camptodactyly**

It is a medical condition involving fixed flexion deformity of the interphalangeal joints.

**Images for this section:**
**Fig. 1:** A: Transverse deficiency: acheiria. B: Transverse deficiency: symbrachydactyly. C: Amniotic band syndrome: acrosyndactyly (amniotic band is seen around digits).

**Fig. 2:** A: Absence of radius and thumb, ulnar bowing B: Absence of distal radius C: Hypoplasia of radius (>2mm shorter than ulna)
Fig. 3: A: Hypoplasia of thumb with strong shortening of the first metacarpal. The carpal bone deficit cannot be evaluated because of the patient's age (< 5 years old). B: Absence of thumb and scaphoid, associated with congenital vertebral fusion in VACTERL syndrome. C: Hypoplasia of thumb. The latter is in the same plane as the other digits. This is a specific sign of Holt-Oram syndrome.
Fig. 4: A: Absence of distal ulna and radial bowing. B: Hypoplasia of ulna and humeroradial synostosis. C: IV-V metacarpal synostosis on the right side and absence of post-axial digits on the left side.
Fig. 5: A: Typical cleft hand in SHFM syndrome (Split Hand Foot Malformation). The third space digit is enlarged and associated with centroaxial syndactyly. B: Atypical cleft hand. Absence of third digit and surnumerary metacarpal bone. C: Atypical cleft hand with postaxial syndactyly and syndactyly of second and third digits. D: Complete carpal coalition in centroaxial deficiency.

Fig. 6: Intercalary deficiency.
**Fig. 7:** Complete or incomplete radioulnar synostosis. Note on the last image irregularities of ulna and radial cortical bones because of the radioulnar synfibrosis.

**Fig. 8:** A: Mesoaxial syndactyly. B: Sympolypdactyly. C: Complete syndactyly.
Fig. 9: Apert syndrome.

Fig. 10: Preaxial polydactylies.
Fig. 11: A and B: Type A postaxial polydactyly. C and D: Type B postaxial polydactyly.
Fig. 12: Polysyndactyly: postaxial polydactyly of the hand and preaxial polydactyly of the foot. Note partial synostosis of the third digit.

Fig. 13: Diagram showing main types of brachydactyly (from Temtamy and Mc Kusik).
Fig. 14: A: Brachydactyly A1. B: Brachydactyly A4. C: Brachydactyly A3. D: Brachydactyly B.
Fig. 15: Different types of brachydactyly C.
Fig. 16: Type E brachydactyly. A: Brachymetacarpy of the fourth digit and Madelung’s deformity in Turner syndrome. B: Brachymetacarpy of the third and fourth digits in Albright Osteodystrophy.
Fig. 17: Sprengel deformity and associated omovertebral bone.
Fig. 18: Congenital pseudarthrosis of the right clavicle.

Fig. 19: Clinodactyly with a delta phalanx in brachydactyly A3.
Conclusion

Despite advance in molecular diagnosis of congenital abnormalities, information provided by clinical and radiological examination is still important before undergoing extensive genetic tests. A systematic analysis of the upper limb will allow correct identification of the abnormality and will serve as a first step in the identification of syndromes and their differential diagnoses. The second step is the integration of associated abnormalities in other organ systems. A genetic opinion for a child with congenital abnormalities would be of great value.

Personal Information

References


