Congenital hand anomalies and reconstruction

The management of congenital hand deformities encompasses the care of the family from infancy, through childhood, to produce a well-adjusted adult who can function physically, socially and psychologically to the highest level that their anatomy will allow. It requires teamwork by all involved for this to be successful.

Congenital hand deformities are frequent, usually isolated anomalies, but may exist as part of a syndrome. The spectrum of upper limb anomalies is broad.

In complex cases, the deformity may not always fit a recognized pattern, requiring bespoke reconstruction, returning to first principles and designing novel solutions. The aim is to create a limb that is functionally optimal but also cosmetically acceptable. Growth increases this challenge since surgery may affect the growth of the skeleton and growth itself may affect the results of surgical intervention.

This review will concentrate on the general management of the more frequent anomalies.

The family

Despite advances in antenatal diagnosis, only a tiny proportion of congenital limb anomalies are diagnosed prenatally. At birth, the family are frequently devastated to find an obvious deformity in their new baby and are fearful of other underlying problems.

The child must be thoroughly examined by a paediatrician for associated defects, some of which, such as cardiac problems, may require more urgent attention than the obvious limb problem. Where multiple anomalies are present, a genetics consultation may help in piecing the picture together. This is essential if any of the family are contemplating further pregnancies.

For several reasons, it is wise to review the child on more than one occasion before surgical intervention. The normal child will demonstrate a developmental change in his/her upper limb capabilities which will be paralleled, to some extent, by the child with limb deformities. Time is needed to evaluate what is likely to benefit the child. The team caring for the child need to gain the trust of the parents. The parents, while undergoing a grief reaction for the loss of the normal child they were expecting, are initially unable to fully comprehend what is being explained to them. The mother experiences guilt. The family want the surgeon to be able to recreate a hand of normal function and appearance. The surgeon needs to explain honestly what is possible to achieve and that growth of the child may necessitate further surgery.

Timing of intervention varies with the condition, other pre-eminent pathology, the size of the child and the philosophy and the abilities of the team caring for the child. Some parents struggle to decide what is best for their child – they should not be pushed into making a decision hurriedly but may be supported and guided through that process by the provision of appropriate information.

As the child matures, he/she will be able to participate more in the decision-making process. Conflict may arise within the family where the child's and parent's wishes do not coincide.

Classification

The internationally recognized classification of congenital hand problems is based on morphology (*Table 1*). It combines groups of disparate conditions together, while others lie in several subcategories in different areas of the classification and approximately 10% of cases do not fit in anywhere. With advances in the understanding of abnormal limb development, this is under revision.

Table 1. International Federation of Societies for Surgery of the Hand classification of congenital hand problems

I	Failure of formation of parts	Transverse	
		Longitudinal	Radial club hand
			Cleft hand
			Ulnar club hand
II	Failure of differentiation of parts		Camptodactyly
			Syndactyly
III	Duplication	Polydactyly	Pre-axial (thumb)
			Central
			Post axial (ulnar)
IV	Overgrowth		Macrodactyly
V	Undergrowth	Hypoplasia	
VI	Constriction ring syndrome		
VII	Generalized skeletal abnormalities		Arthrogryposis

From Swanson (1976)

Miss Gillian D Smith *is Consultant Plastic, Reconstructive and Hand Surgeon in the Department of Plastic and Reconstructive Surgery, Great Ormond Street Hospital for Children, London WC1N 3JH (Smithg3@gosh.nhs.uk)*

Aetiology

The majority of conditions do not have their aetiology firmly established. An increasing number are recognized to have a genetic basis, many as spontaneous new mutations. The thalidomide tragedy of the 1960s identified a single teratogen associated with limb anomalies but this situation is rare and few drugs or environmental factors have been implicated.

Incidence

The incidence of each condition varies widely in the literature as a result of poor initial reporting of limb anomalies and different frequencies within different population groups.

Syndactyly (webbed digits)

Syndactyly is probably the commonest congenital hand abnormality. It occurs in 1:1000 live births. It may be unilateral or bilateral, complete or incomplete (not extending to the distal phalanx), simple involving skin only (*Figure 1*), or complex with bony connections (*Figure 2*). It may be part of a syndrome, genetically inherited or isolated and sporadic. It may have associated extra digits either as a complete ray, concealed within a digit or in its most complex form as a confusing jumble of bones spanning several rays. Acrosyndactyly, with distal connections between the digits but proximally the webs are present, is seen chiefly as part of constriction ring syndrome.

Syndactyly may affect fingers and/or toes. The commonest form of syndactyly is an incomplete second web syndactyly in the foot, and third web syndactyly in the hand. The first web is the least commonly affected. Syndactyly of border digits (thumb and little finger) needs to be released by the age of 6 months to allow adequate longitudinal growth of the fingers and independent thumb function.

An absence of skin creases in the digits suggests a failure of active motion. Some parents erroneously believe that motion will be restored with syndactyly release, especially where passive joint motion remains – this fallacy must be addressed before surgery.

Neurovascular anomalies within the web are increasingly likely with increasing complexity of the syndactyly, occasionally making separation impossible.

There are many techniques described to correct syndactyly but all are based on using flap reconstruction to form a new web, avoiding straight line scars across the web or crossing the flexor surface of the joints and importing new tissue to replace the deficit, usually in the form of full thickness skin grafts.

Figure 1. Simple syndactyly.

Symbrachydactyly (short webbed fingers)

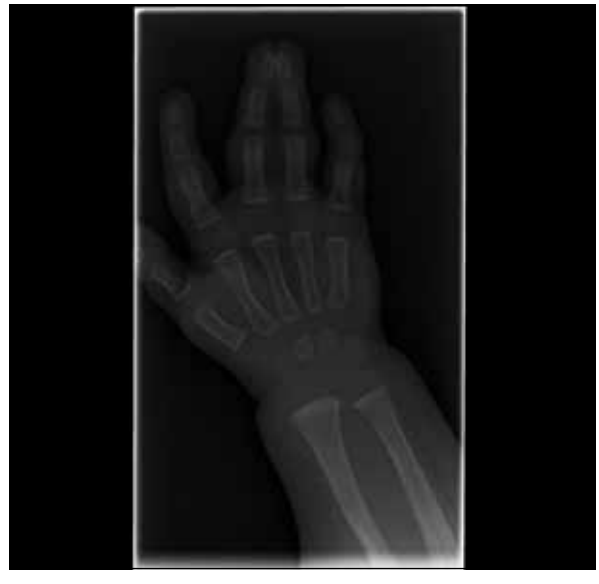
This abnormality is unilateral, sporadic and has an estimated incidence of between 1:10 000 and 1:30 000 live births. It may be associated with Poland's syndrome where the patient presents with loss of the sternal head of the pectoralis major, chest wall deformity, nipple asymmetry with later breast asymmetry, abnormalities of the latissimus dorsi and serratus anterior, and gradually increasing hypoplasia of the limb progressing from proximally to distally.

Symbrachydactyly constitutes a spectrum of disorders with the least severe manifesting as a slightly smaller hand than on the contralateral side with smaller or absent middle phalanges. The most severely affected hand will have small metacarpals but no digits, only residual nubbins. Treatment depends on the individual anomaly but most patients are functionally excellent since they have one normal upper limb. For the most mildly affected, with short fingers, no treatment is necessary whereas for the most severe, prostheses or a double microsurgical free toe transfer will provide the most useful assisting hand.

Camptodactyly

Camptodactyly, meaning 'bent finger', describes a flexion deformity at the proximal interphalangeal joint of a single or multi digits. The little finger is most frequently

Figure 2. Complex syndactyly.



affected and bilateral involvement is more common than unilateral (*Figures 3, 5*). The digits have full flexion but are unable to be fully actively extended and, in the later stages, unable to be passively extended. Where camptodactyly affects multiple digits on each hand, it is more likely to be associated with a syndrome and is less likely to respond to treatment. Two groups of patients are affected – infants and teenagers. Infants in the first 2 years of life have an equal sex incidence of this condition. In teenagers, girls are more frequently affected.

The mainstay of treatment is passive stretches and splintage until full extension is obtained and maintained. Radiographs may reveal a joint deformity with flattening of the proximal phalangeal head.

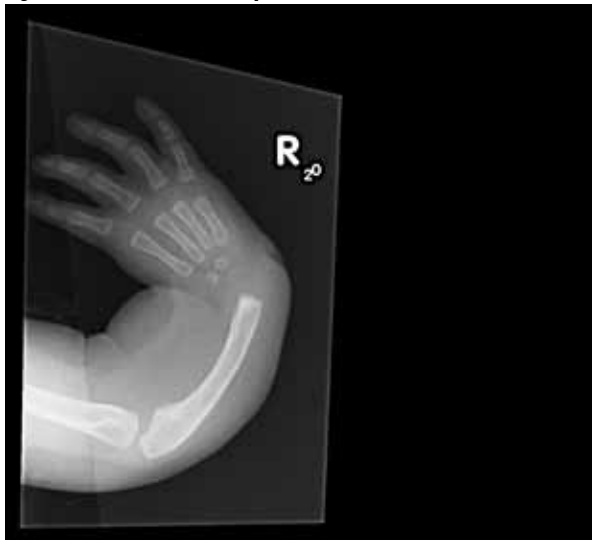
Surgery can be deceptively complex as all the structures on the volar aspect of the joint may be short. Surgery is thought to only be worthwhile in those with a deformity of >60° but, even in these patients, surgical results are not encouraging. There is a risk of devascularization of the digit since the vessels may not tolerate full extension. Only a minority of patients obtain both full active flexion and full active extension postoperatively.

Radial longitudinal dysplasia (radial club hand)

Here the hand is found to be radially deviated, unstable, flexed, and pronated on the forearm, as a result of varying degrees of hypoplasia or aplasia affecting the soft tissues and the skeletal elements on the radial side of the distal forearm and hand.

It may be unilateral or bilateral but is rarely symmetrical. Its estimated incidence is between 1 in 30 000 (Flatt, 1994) and 1 in 100 000 live births (Birch-Jensen, 1949). Bayne and Klug divided radial club hand into four groups according to the degree of radial hypoplasia: type I – short distal radius, type II – hypoplastic radius, type III – partial radial aplasia and type IV – radial aplasia. Type IV is the most frequent type undergoing surgery.

Figure 3. Radial club hand – pre-centralization.



Aetiology remains uncertain; genetic factors play a role in syndromic cases. The myriad of reported syndromes associated with this condition are mainly best left to the specialist geneticist. Some require early exclusion by the generalist including those associated with haematological conditions, such as Fanconi's anaemia and thrombocytopenia absent radius syndrome, cardiac and renal defects.

Fanconi's anaemia is an autosomal recessive condition which produces a pancytopenia, fatal in the absence of bone marrow transplant. These patients might never be appropriate for surgical intervention.

Other conditions may necessitate delaying surgery. In thrombocytopenia absent radius syndrome, the patient has bilateral radial club hand with short forearms but

Figure 4. Distraction in radial club hand.

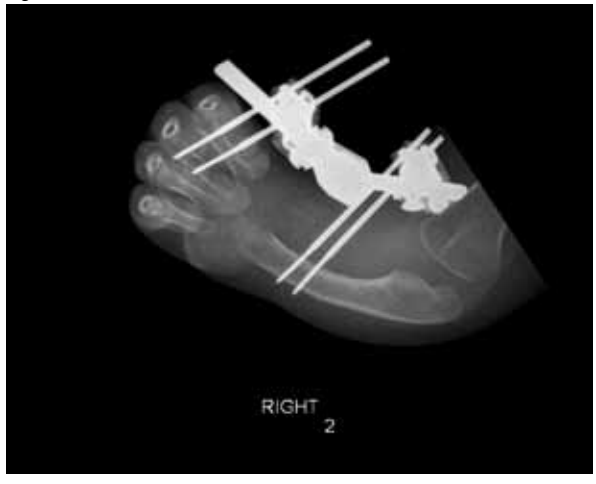
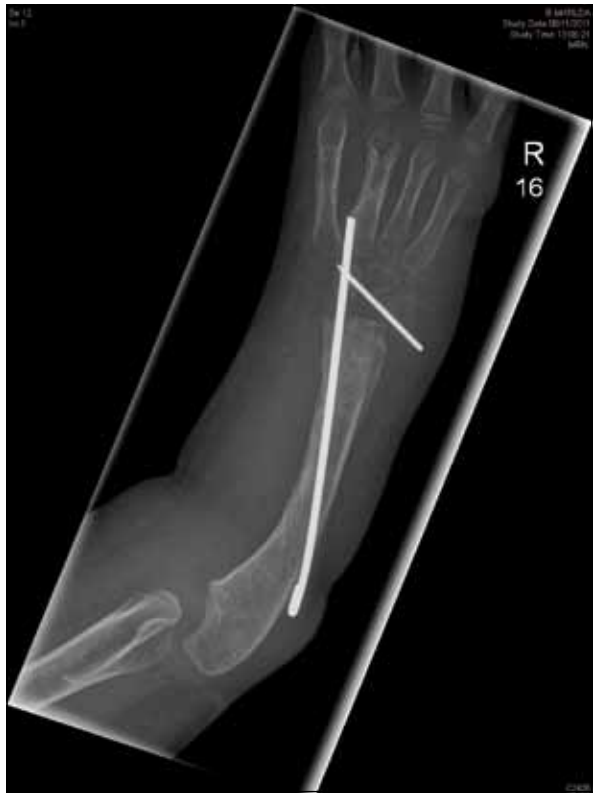


Figure 5. Radial club hand post centralization.



thumbs present and a platelet count which improves with age until it approaches normal at around 5 years.

Cardiac defects, commonly atrial and ventricular septal defects, in association with radial ray deficiencies are seen in Holt–Oram syndrome. This is an autosomal dominant condition affecting T-box (TBX) genes. Other family members may be affected and undiagnosed.

Patients with VACTERL syndrome form the largest syndromic group with radial club hand. They have a minimum of three of the following: Vertebral abnormalities, Anorectal, Cardiac, Tracheo-oEsophageal, Renal, Limb anomalies and a Single umbilical artery. Their radial deficiency is often severe with stiffness and camptodactyly of multiple digits.

In radial club hand, the upper arm is short, the elbow may be stiff, pronation and supination are limited or completely absent. The forearm is short, approximately 60% of normal length, and the ulna often bowed. The radius is always deficient distally; if present, it may also be abnormal proximally. The thumb has varying degrees of hypoplasia or is absent. The digits are increasingly deficient moving from the ulnar to radial side of the hand.

Treatment is aimed at functional improvement to correct the wrist position while maintaining motion and growth capacity. It begins with physiotherapy and splintage by the parents and hand therapist stretching the wrist to bring it to neutral. If this is easily achieved, then a tendon transfer of the dorsoradial muscle mass into the extensor carpi ulnaris may rebalance the wrist.

In patients with types III and IV radial club hand, soft tissue distraction using an external fixator allows gradual stretching of the soft tissues to simplify the subsequent wrist rebalancing surgery (*Figures 3* and *4*). Following distraction the hand needs to be balanced on the end of the ulna by means of soft tissue release, tendon transfers and/or bony resection. In radialization, the ulna is placed in line with the second metacarpal and the stability is dependent on the quality of the tendon transfer. In centralization, alignment is with the third metacarpal and a carpal slot assists the tendon transfer in obtaining stabilization (*Figure 5*).

Table 2. Blauth classification of thumb hypoplasia

Blauth grade	Soft tissue anomalies	Skeletal anomalies	
I	Minor hypoplasia of entire thumb	Normal	
II	Adduction contracture of first web, median innervated intrinsic muscles absent	Lax ulnar collateral ligament of metacarpophalangeal joint, slim intact skeleton	
III	All of above and extrinsic muscle anomalies, absent intrinsic muscles	Partial aplasia of metacarpal base	A: Unstable metacarpophalangeal joint, carpometacarpal joint present
			B: Absent carpometacarpal joint
IV	Floating thumb, no muscles	Absent metacarpal	
V	Absent thumb	Nil	

From Blauth (1967)

Thumb hypoplasia (underdeveloped thumb)

Thumb hypoplasia is a form of radial ray deficiency. The Blauth classification (*Table 2*) subdivides thumb hypoplasia into five groups with gradually increasing degrees of skeletal abnormality. In reality, any grade of thumb hypoplasia can have either or both intrinsic muscle and extrinsic tendon abnormalities which may affect its functional abilities to differing degrees.

Grade I thumbs rarely need treatment. Grade II and IIIA require reconstruction with first web release, stabilization of the metacarpophalangeal joint and tendon transfers for opposition. Grade IIIB thumbs and greater are best treated by amputation and pollicization of the index finger. Attempts at reconstruction have produced functionally and aesthetically inferior results compared to pollicization.

Pollicization, the creation of a thumb from an index finger, is a technically demanding procedure, highly dependent for its results on both the quality of the index finger and the surgeon's abilities. The best results will still produce a thumb that is only half the strength of the normal thumb but which allows opposition and grasp of large objects.

Cleft hand

The classical cleft hand is an autosomal dominant condition affecting both hands and feet and has an estimated incidence of 1:90 000 births (Birch-Jensen, 1949). It may be associated with other anomalies such as cleft lip and palate, talipes, ventricular septal defect, Fallot's tetralogy or be part of ectrodactyly–ectodermal dysplasia clefting syndrome. Patients usually have excellent function but aesthetically the appearance is a disaster. The surgeon must be wary of interfering with function to improve the aesthetic appearance.

The cleft involves absence of one or more central rays and may include proximal phalanges that lie transversely in the web and will cause the cleft to widen with growth. There is variable narrowing of the first web space (*Figure 6*).

The aims of surgery are to free the thumb, recreate a first web space by importing tissue from the central cleft, and to close the central cleft, reconstructing the transverse metacarpal ligament across the cleft and reconstructing the central web with a distally based flap. If there is a bone lying transversely across the cleft, this should be removed.

Ulnar club hand

This is the rarest of the longitudinal deficiencies with an incidence of 1:100 000 live births and is frequently bilateral, affecting the whole of the upper limb. Other associated anomalies are largely musculoskeletal. The wrist is relatively stable but the elbow is deficient or absent. There are often absent digits on both the radial and the ulnar side of the hand, hypoplasia of the thumb and

simple or complex syndactyly in those digits present (*Figure 7*). Radial head dislocation is common but usually asymptomatic.

Treatment is largely aimed at addressing the hand abnormalities.

Macrodactyly (large digit)

In macrodactyly, one or more digits, usually in the median nerve distribution, are larger than normal and can grow disproportionately – the latter usually come to amputation whereas repetitive debulking procedures ameliorates the less progressive type.

Trigger fingers

Congenital trigger digits are probably not present at birth but likely to be developmental. Trigger thumbs are fourteen times more common than trigger fingers. They may present with a nodule in the palm (Notta's nodule) at the base of the A1 pulley. Trigger fingers may present with triggering but paediatric trigger thumbs usually present with a flexion deformity (*Figure 8*) which may be misdiagnosed by the unwary as a dislocation.

Trigger thumbs are known to resolve but the frequency and timing with which this occurs are disputed. Initially, stretching exercises are used and sometimes splintage. Traditional teaching suggests surgical intervention with release of the A1 pulley before the age of 3 years. This has excellent results with few complications. Correction of trigger fingers usually does not respond to division of the A1 pulley alone and the best results have been reported when one slip of flexor digitorum superficialis is sacrificed concomitantly.

Duplication (extra digits)

Polydactyly competes with syndactyly for the most frequent congenital hand disorder.

Ulnar polydactyly (extra little finger)

Extra digits on the little finger have a marked racial preponderance for the Afro-Caribbean population, usually with autosomal dominant inheritance. Syndromic associations occur, particularly where the extra digit is more substantial (*Figure 9*).

Figure 6. Cleft hand.

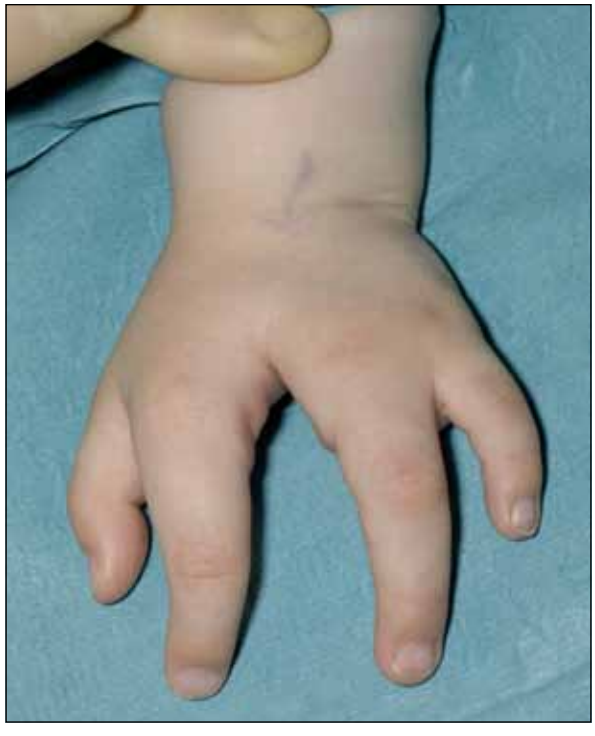
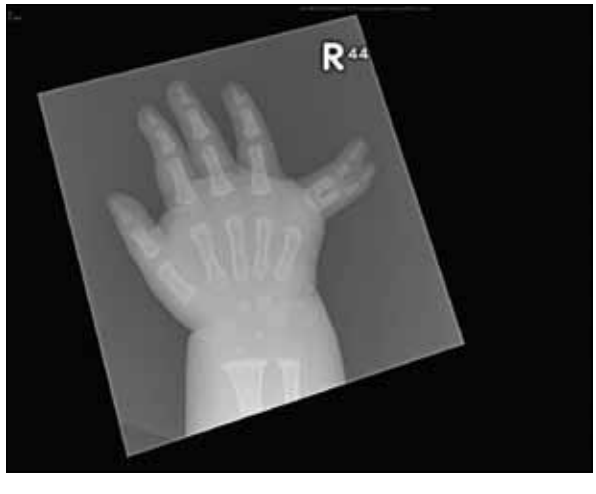


Figure 7. Ulnar club hand.

Figure 8. Trigger thumb.

Figure 9. Ulnar polydactyly.



Stelling (1963) divides ulnar polydactyly into three groups:

Type I Soft tissue mass without skeletal structure
Type II Digit with all normal components and articulating with a normal or bifid metacarpal or phalanx
Type III Complete digit with metacarpal.

Type I can be treated by formal removal in the first 8 weeks of life under local anaesthetic. Type II may require partial resection of the metacarpal head and reconstruction of the metacarpophalangeal joint and will require reattachment of abductor digiti minimi tendon. Type III requires a ray amputation.

Central polydactyly (extra central digits)

Central polydactyly, although rare, is accompanied by varying degrees of syndactyly. Polysyndactyly, where it produces polysyndactyly of the fifth ray of the foot with a suppressed polysyndactyly of the fourth ray of the hand, is known to be associated with abnormalities in the Homeobox D13 gene. Bony and neurovascular anomalies, symphalangism and tendon abnormalities make central duplications tricky to correct. Results are functionally and aesthetically less pleasing than other forms.

Radial polydactyly (thumb duplication)

Thumb duplication, classified by Wassel (1969) (*Table 3*), is rarely part of a syndrome and is, with the exception of the type VII triphalangeal thumbs, usually unilateral. Each of the two thumbs is smaller and narrower than the contralateral thumb. The duplicates may be symmetrical or not – asymmetry makes decision making on which thumb to keep more straightforward (*Figure 10*). Where the deficiency is symmetrical, a procedure sharing parts of both thumbs may be needed. In types I and II with similar sized thumbs, the Bilhaut–Cloquet procedure, where the thumbs are joined in a side-to-side manner, is often recommended, although rarely performed because of issues with persistent nail and epiphyseal deformity.

Type IV (*Figure 11*) is seen most frequently and here there is a bifid metacarpal head. The intrinsic muscles must be detached, anomalous tendinous connections (such as pollex abductus) divided, tendon insertions corrected, the metacarpal head shaved to leave a single articular surface and the collateral ligament reconstructed to the metacarpophalangeal joint with reattachment of the intrinsic muscles and enlargement of the first web space where required.

Type VII thumbs may require a formal pollicization.

Table 3. Wassel's classification of thumb duplication

Type I	Bifid distal phalanx
Type II	Duplicated distal phalanx
Type III	Bifid proximal phalanx
Type IV	Duplicated proximal phalanx
Type V	Bifid metacarpal
Type VI	Duplicated metacarpal
Type VII	Triphalangia, in either or both of the thumbs

From Wassel (1969)

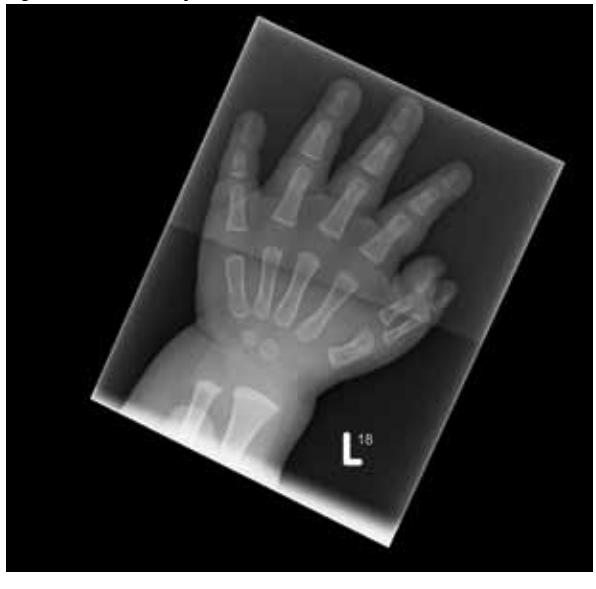
Arthrogryposis

The term arthrogryposis refers to a collection of conditions where there are multiple joint contractures. The classical form, arthrogryposis multiplex congenita, consists of joint contractures and muscle wasting from birth without evidence of any progressive neuromuscular disease. Its aetiology is not firmly established although a viral illness affecting the anterior horn cells in the spinal cord is thought probable. The limbs have few skin creases, tight muscles and joint capsular contractures in a characteristic form: the shoulders internally rotated, the elbows extended, forearms pronated and the wrists flexed and ulnar deviated with the digits flexed and the thumb lying within the palm. The lower limbs show talipes equinovarus and dislocated knees and hips.

When operating on these children, the principles that must be applied are to avoid loss of the arc of motion of joints, to increase passive range primarily as that may improve function by the use of trick manoeuvres, to

Figure 10. Thumb duplication.

Figure 11. Thumb duplication.



maintain bimanual function as neither hand will function well independently, to position the limbs in front of the body at the level of the desktop and to maintain function for assisting lower limb needs for transfers or the use of walking aids.

The single most useful function the surgeon can gain is passive elbow flexion. Attempts at obtaining active flexion may risk producing an elbow contracture and limiting ability to transfer. The most useful procedure for the wrist is a wedge resection of the carpus with tendon transfers to augment extension. The thumb exhibits an adduction contracture, laxity of the ulnar collateral ligament and an absence of abductor pollicis brevis, best served with first web release and metacarpophalangeal joint epiphysis-sparing fusion.

Children with classical arthrogryposis have an intelligence quotient above average compared to their peer group, so providing sufficient function to allow them to feed themselves, and to transfer into and operate a wheelchair and a computer keyboard may allow them to lead a relatively normal life for someone with such generalized impairments.

There are a group of arthrogryposes which comprise more distal involvement with contractures and intrinsic muscle dysfunction confined to the hands – these are a more varied group but a combination of splintage with stretches and surgery to the thumb where required is often sufficient to maintain reasonable function.

Ring constriction syndrome

This has an incidence of 1:15 000 live births and is associated with talipes equinovarus, cleft lip and palate and other defects in the child and oligohydramnios in the mother. Whether it is the result of extrinsic constriction from amniotic bands or an intrinsic defect in the germ cell layer is debated. Patients present with a variable combination of simple constriction rings, amputation of digits, acrosyndactyly and occasionally constriction rings associated with distal lymphoedema. Where there is lymphoedema distally, urgent release of the ring may be required within the first few days of life. Early surgery may be required to release acrosyndactyly and allow individual digits to grow independently (*Figure 12*). Excision of rings is often to the level of the deep fascia and often only one ring can be excised on a digit at a time if there is to be sufficient remaining tissue for closure. Where multiple amputations occur, these may be candidates for distraction augmentation manoplasty or for free toe transfer to create better grip and digital length.

Figure 12. Constriction ring syndrome.

Microsurgery

Microsurgery has an important and emergent place in the treatment of the congenital hand deformity. Free flaps to create an adequate first web, free vascularized muscle transfers to allow restoration of flexion or extension after compartment syndrome of the forearm musculature and free vascularized joint transfers have added to the armamentarium available for treatment.

However, free toe transfer used to reconstruct the thumb and absent digits with good functional results in the appropriate patient has been where microsurgery has shown its greatest benefit. The aesthetic results of this surgery have improved by reducing the pulps of the toes as a secondary procedure. The donor site from harvesting a single second toe from each foot leaves no functional problems.

Conclusions

Congenital hand anomalies remains a highly specialized and challenging field where the needs of the child are addressed planning for the function of the future adult. Advances in fields such as prosthetics, gene manipulation and stem cell technology may hold the key to future improvements. BJHM

Conflict of interest: none.

Birch-Jensen A (1949) *Congenital deformities of the Upper Extremities.* Andelsbogtrykkeriet i Odense & Det danske Forlag, Odense

Blauth W (1967) Der hypoplastische Daumen. *Arch Orthop Unfall-Chirurgie* **62**: 225–46

Flatt AE (1994) Radial clubhand. In: Flatt AE, ed. *The care of congenital hand anomalies.* 2nd edn. Quality Medical Publishing, St. Louis

Stelling F (1963) The upper extremity. In: Fergusian AB, ed. *Orthopedic Surgery in Infancy and Childhood.* Williams and Wilkins, Baltimore: 304–8

Swanson AB (1976) A classification for congenital limb malformation. *J Hand Surg* **1:** 8–22

Wassel HD (1969) The results of surgery for polydactyly of the thumb. *Clin Orthop* **64**: 175–93

Further reading

Smith PJ (2002) *Lister's The Hand, Diagnosis and Indications.* 4th edn. Churchill-Livingstone, London

KEY POINTS

- The care of the congenital hand patient needs to be multidisciplinary and holistic involving paediatrician, surgeon, prosthetist, therapists and parents.
- Care is individualized to the condition and to the patient and family.
- Follow up for most conditions is until adulthood.
- The aim of care is to produce a well-rounded functional adult with the best cosmetic and functional upper limbs that the condition allows.