The principles of management of congenital anomalies of the upper limb

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Abstract
Management of congenital anomalies of the upper limb is reviewed with reference to classification and aetiology, incidence, diagnosis before birth, broad principles of treatment, timing of x rays and scans, functional aims, cosmetic appearance, counselling of parents, therapists, scars, skin grafts, growth, and timing of surgery. Notes on 11 congenital hand conditions are given.

There are six current textbooks referenced. 1–6 Each is an excellent starting point for reading on this subject; together they provide in depth coverage and details of surgical techniques. There is little consistency in the noun used. Deformity, anomalies, malformations, are all used, often in the same text and also in an attempt to get away from stigmatising nouns, Graham Lister uses “differences”.6

Classification and aetiology
The currently used classification of congenital anomalies of the upper limb is based on that of Swanson modified by the Congenital Malformations Committee of the International Federation of Societies for Surgery of the Hand (IFSSH) in 1983.1–4 7

I. Failure of formation of parts (arrest of development)
A. Transverse arrest (common levels are upper third of forearm, wrist, metacarpal, phalangeal)
B. Longitudinal arrest (including phocomelia, radial/ulnar club hands, typical cleft hand, atypical cleft hand otherwise referred to as part of the spectrum of symbrachydactyly)

II. Failure of differentiation of parts
A. Soft tissue involvement
B. Skeletal involvement
C. Congenital tumorous conditions (includes radio-ulnar synostosis, symphalangism (stiff proximal interphalangeal joints (PIP)s with short phalanges), camptodactyly, arthrogryposis, syndactyly)

III. Duplication
IV. Overgrowth
V. Undergrowth (thumb hypoplasia, Madelung’s deformity (abnormal distal radial growth)
VI. Congenital constriction band syndrome
VII. Generalised skeletal abnormalities.

These seven groups have the attraction of grouping together what seem at first glance to be similar conditions. The more closely each group is analysed, however, the more of a mixed bag of conditions they seem to be, and the more one can question whether they should be grouped together. For instance, typical cleft hand, although it has absent parts to the centre of the hand, does not have any hypoplastic parts, whereas symbrachydactyly is a whole range of degrees of absence and hypoplasia. The known inheritance also varies within groups from dominant genes in typical cleft hand, in Madelung’s and some syndactyly families,9 to known associations in the Vater cases, to conditions with no known hereditary element.

So this classification, although giving some insight into the embryological problem, contains classification uncertainties. Others, such as Ogino,9 have proposed improvements to address some of the uncertainties, but they persist. Such classifications are of little practical value in the everyday management of congenital hand malformations. Each case should be analysed in its own right, the malformations described, and treatment planned on this analysis.

There is growing insight into the early development of the limb and its gene control.10 11 As this understanding unfolds we should have a classification which would be more secure. A genetic opinion on a child with congenital anomalies is of great value, both to the treating team and to the parents, and should always be sought.

Incidence
Estimates of incidence from different countries are discussed in the standard textbooks. One set of figures comes from the Congenital Malformations Committee of the International Federation of Societies for Surgery of the Hand, published by Lamb in 1982.7 14 They give the minimum estimate of cases per 10 000 total births. Their total is 22.91. Of this all types of polydactyly together were 9.5, all types of transverse failure of formation at hand/finger level 5.8, syndactyly 1.5, hypoplasia 1.3, radial club hand 0.8, ring constriction 0.6, and amelia 0.2. Compare this with about 14 clefts per 10 000 births.

Diagnosis before birth
Antenatal diagnosis by ultrasound is increasingly possible, particularly in cases of aplasia. This will prepare the parents for the deficiency before birth.
Broad principles of treatment

The hand surgeon’s aim is to achieve the maximum upper limb function possible to suit each individual patient, always bearing in mind the importance of cosmetic appearance. Physiotherapy and occupational therapy splintage are invaluable in many conditions and therapy should start in the neonatal period. This treatment in not invasive and can run concurrently with the treatment of other conditions. Early referral to the hand surgeon and specialist therapist is important.

Children with congenital anomalies of the upper limb must be thoroughly assessed, both from their general physical (including haematology in the case of radial club hand) and mental development. The specialist hand surgeon planning the treatment of congenital hand problems has to be very much aware that he is providing only part of the developmental needs of the child.

The upper limb anomaly may well be part of a wider anomaly, for instance the VATER association (vertebral, anal, tracheo-oesophageal, radial club hand), sometimes expanded to VACTERL to recognise the associated cardiac and renal anomalies that occur. Radial club hand is an associated cardiac septal defect in the dominantly inherited Holt-Oram syndrome. Radial club hand can be associated with rare haematological conditions: thrombocytopenia in TAR syndrome and pancytopenia of Fanconi’s anaemia. Apert’s syndrome is the association of craniofacial anomalies with miten hands.

Some operations need to be done in the first few months of age, but in the majority of congenital conditions, surgery is best undertaken between 6 and 18 months. Some operations are long; for instance toe to hand transfers take five to 10 hours and are usually performed between 2 and 4 years.

The treatment must be integrated with the management of the child’s other medical and surgical problems and indeed the child’s normal developmental need (schooling for instance). The safe management of operations is very much dependent on the screening of children by the different medical services (general practitioner, community services, hospital paediatric services, and other hospital specialities) to ensure that the child is thriving and to diagnose or exclude other medical conditions.

The hand surgeon cannot assume that the different medical services know about the planned hand surgery. The hand surgeon must actively check that everyone is in agreement and that the timing of the surgery is optimal and the child fit.

Above all talk to the child’s mother and father. If language is a problem it is essential to have a consultation with a fluent interpreter. The mother with older children will know if this child is well, feeding well, growing, and meeting its milestones. The mother with her first baby may not know what is normal and needs time and careful questioning and counselling.

The anaesthetist must be experienced both in anaesthetising infants and young children and also in the anaesthetic requirements of modern hand and microsurgical operations. They should see the child preoperatively, with plenty of time to talk to the parents and the nurse who has been looking after the child. The day before is the best time but with the increasing demand for day surgery and reducing the number of nights in hospital, the anaesthetist often does not see the child until the day of surgery. The child needs one and a half hours for the Emla cream to take effect before going to theatre.

Modern hand surgical procedures on children and on adults share the common need for specialist nursing teams, specialist equipment needed in microsurgery, bone fixation, and imaging the skeleton. Long operating times are often needed. The surgeon needs to be practiced in all aspect of hand surgery including microsurgery. Paediatric hand surgery is not for the occasional operator. It must take place in the correct environment for the management of children, with all the paediatric support services that are required. It is important to intimate up the paediatric needs with surgical needs for a successful outcome. In some cities the main paediatric facilities and the surgical units where hand and microsurgery are regularly practiced are not on the same campus. Hence one or other has to be duplicated at the other site and the two disciplines must understand each other’s difficulties.

The timing of x rays and scans

X Rays in the neonatal period give some idea of the bony anomaly but are not as useful as x rays at a few months of age just prior to surgery. So take x rays if you have a reason to, but otherwise leave them for later. The new imaging machines that are available to hand surgeons for use in the operating room and outpatients are of such low dose of radiation that they are very useful in children who will need many x rays over the years.

Functional aims

Sensation is paramount. A part with no sensation is not used and does not grow. Congenital anomalies will all have normal sensation. This is different to some obstetric brachial palsy patients or injured patients. Any reconstructive surgery that involves a nerve anastomosis will only be successful if there is a good level of sensory return. Nerve recovery in very young children is considerably better than in older children and is a strong reason for very early surgery, as for instance in toe to hand transfers.

The patient’s general and mental capacity and its prognosis must be assessed. A child with limited mental capacity still needs hand function, bimanual if possible, but the aims might be more limited. For instance in the miten hand of Apert’s syndrome we might be content with a tripod hand (thumb, central finger mass, and little finger) instead of separating all four fingers. This saves a considerable amount of additional surgery that is required to bringing only a limited function gain that the child may not be able to utilise.

In unilateral malformation with a normal hand on the other side it is often felt that there may be less need to treat than in the bilateral deficiency. Flatt says that 90% of the tasks of
daily living can be performed with one hand. This may be so but the child is best served by maximising hand function.

It is very important to assess any spinal or leg problems affecting mobility, and the prognosis of the mobility needs to be known. The upper limbs may be needed to use walking aids and be needed for transfer from bed to chair. In these children at least one elbow stable in extension is needed. This can be important in planning the treatment, for instance in cases of arthrogryposis.

Most other functions require a flexed elbow, preferably with some range of movement around the flexed position and with some shoulder movement. This is true for feeding, for toilet needs, and for many functions like dressing, playing, and writing.

The hand requires as good a shoulder and elbow movement as possible to position the hand. The functional aims can be planned broadly in the following descending order of aims.

**PREHENSILE ABILITY WITH AN OPPOSABLE THUMB**

The normal hand can oppose the thumb to the four finger pulps and manipulate objects with great dexterity. The finest of movements uses the most distal joints. It requires both the extrinsic muscles and the intrinsic muscles to control the interphalangeal joints and give the digits their dexterity.

Thumb to two finger tripod pinch still gives the fine ability to manipulate objects (fig 1). Thumb to single finger pulp pinch, usually index, gives valuable precision and is the basic reconstructive aim of hands with digit aplasia (fig 2). Cruder thumb to single finger pinch, often ring or little finger, gives opposition and the ability for holding objects of a certain size, even though the two digits do not have enough movement to touch. This is the reconstructive aim of many toe transfers where there is no normal thumb (fig 3).

Key pinch is thumb against the side of the finger or fingers. This lacks the sensation and precision of pulp to pulp pinch but is a strong and useful form of pinch. It is the reconstructive aim in hands where the thumb has only limited ability to abduct and oppose and where the fingers lack much movement.

**NON-PREHENSILE FUNCTION**

*Individual digit function with sideways movement* is restored in syndactyly release.

*Finger grip with some power*—This is an important power function of the human hand, requiring finger length and interphalangeal movement. It cannot usually be effectively reconstructed in the anomalous hand.

*Palm as sensate paddle with wrist movement* is a useful function. It might be possible to give prehension by two toe transfers (fig 3); otherwise function can be augmented by opposable prosthetic aids, each aimed at specific functions such as holding, or in later years, driving.

*A small piece of proximal palm with wrist movement*—This in itself is probably more useful than a prosthesis because of its sensation. Its function is increased with prosthetic aids.

*Transverse deficiencies of the forearm with elbow movement* act as a hook to hold objects with straps or to fit a prosthesis.

*Thoraco-humeral pinch* is a very basic holding function of the short arm.

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**Figure 1** Apert’s mitten hand. (A) Preoperative. (B) Postoperative; little finger and thumb separated to give tripod pinch.
Cosmetic appearance
The first aim of treatment is function, but the cosmetic appearance comes a very close second. One important aspect of appearance which should be understood is that appearance is more enhanced by a hand that moves and gestures naturally and by the presence of an opposable digit than by the number, shape, and length of the digits. The hand that is immobile is strikingly noticeable. The hand that moves normally is accepted. Who counts Mickey Mouse’s fingers?

The radial club hand is a very noticeable deformity but, because it points towards the mouth, it is very functional. Straightening the hand makes it look better and the forearm appear longer, but may not increase function if the wrist loses movement, particularly if the elbow is stiff.

The cleft hand is a very noticeable anomaly but very functional. The cleft can be narrowed or eliminated but this will not improve function.

Prosthesis
Always keep in mind the results of prosthesis in proximal deficiencies. Prosthetics just for cosmetic appearance at the expense of covering up sensation are seldom valuable, and a young child will discard them. Cases of proximal aplasia, of which aplasia at the junction of the upper third and middle third of the forearm is the most common, are referred to specialist limb fitters in the first year of life. A simple prosthesis is often fitted in the first or second year, progressing to myoelectric prosthesis during the first decade whenever possible.

Counselling the parents
The first consultation over a child with a major anomaly of the upper limb can be difficult for both the parents and for the surgeon. Often parents do not realise the extent of the malformation and how it will or will not develop, or they are expecting a better result from treatment than the child can get. They may have been given false hope or wrong information by well meaning but ill informed medical personnel. It falls on the specialist consultant to outline the anomaly, and to predict the future outcome and the treatment plan that is required to achieve it. This can come as a blunt truth. It is natural for parents to want a perfect child, and the realisation of the final outcome and its effect on the child’s future can be very difficult for parents to accept.

Helping the parents to understand what is involved in the surgical treatment requires time and patience. There are many aspects for the parents to understand: several operations may be necessary, scars may be produced on visible areas like the dorsum of the hand, and there may be other scars produced at remote sites for skin grafts. What will the final result look like? This can be particularly difficult with some modern reconstructive procedures that are seen as “invasive” or possibly “destructive” and
that sacrifice another normal piece of anatomy like a toe or index finger. Also parents must understand there can be a failure rate in surgery, albeit very small.

A prime example of this is pollicisation. The modern operation of pollicisation was developed and refined by Buck-Gramko and is widely recognised as an excellent reconstruction for hands with thumb aplasia and no trapezium. It is a difficult and very specialised operation but the results are excellent. It converts the index finger into a thumb, producing a prehensile hand with a thumb and three fingers (fig 4). The function is excellent and so is the appearance, but many parents find it difficult to agree to the loss of the index finger.

Toe to hand transfers to reconstruct a thumb or finger aplasia pose a difficult decision for parents. It is invariably the second toe that is used and these very much look like a second toe when transferred to the hand, although growth and a pulp-plasty do modify this to some extent. The functional gain however is enormous. With improved function and movement the cosmetic appearance is improved.

Much time needs to be spent helping parents to understand the benefits of surgery as well as the operations themselves and the appearances of wounds. This is done with the help of videos and photos and above all by showing them other patients who have had the treatment and letting the parents talk.

Specialist counsellors or therapists with time to spend are invaluable. The self help groups (in the UK: Reach, Erbs Palsy Group, Arthrogryphosis Group) fulfil a vital role in all the above and the parents should be strongly encouraged to talk to them.

Many parents may need second opinions before they can come to terms with the treatment options. This puts pressure on NHS resources, but in the long run, if the parents and child understand the options better preoperatively, the surgery and postoperative course will be much smoother.

Information is increasingly available on the Internet. This may be valuable, but there are examples of parents chasing unrealistic surgical promises in other countries, which have not been validated. They should carefully talk through such offers with their first or second specialist opinions and with their paediatrician.

Therapists

In hand units the division between physical therapy and occupational therapy is slowly breaking down so that physiotherapists make splints and vice versa. Again paediatric expertise is essential but so also is adult hand experience, in order to give the therapist a broad framework of the long term outcomes of treating upper limb problems.

A considerable amount of therapy is needed, the keystones being stretching (every nappy change is a good time) and splintage. It is essential to have parental understanding and input. Community therapy services are very valuable but close communication with the hospital services is needed to ensure shared objectives and continuity of care, and to help each other out at times of staff shortages.

In some conditions therapy needs to start in the neonatal period, for instance radial club hand, flexed proximal interphalangeal joints lacking extension, and the “wind blown” conditions where the thumb or fingers lack extension. Often great improvements can be made.

Scars

Scars need to be minimised and hidden wherever possible. The scars are disturbing to parents and children so their natural history must be understood. Many children however are more comfortable with a scar which can be put down to an operation or an injury than a congenital anomaly which they find harder to accept.

Scars in young people are typically red and often raised (hypertrophic). With time (which can be five to ten years), the hypertrophy settles to flat white papery scars. Compression
therapy can accelerate this resolution and is very worthwhile. This is done with customised elasticised garments, gloves, and armbands, which exert steady pressure. These need to be remade frequently as the child grows. They are made in the therapy departments of most large plastic units.

Scars do not grow as much as normal skin; they have to be placed so as not to cause contractions and restrict growth.

**Skin grafts and flaps**

Many congenital hand operations require the use of skin grafts to resurface parts of the digits. Syndactyly needs large grafts. In all areas except for small areas on the dorsum of the hand full thickness grafts are used. Full thickness grafts have the advantage over split skin grafts. They do not contract and they grow with the child without restricting growth, providing that the scars at the edge of the grafts are well placed. They are thick and have a more predictable and even colour. They are taken from the groin crease.

Flaps are thick pieces of tissue with their own blood supply. They provide the best tissue for growth and cosmetic appearance and the best cover when future operations are needed.

**Growth**

Most congenital malformations grow in the same proportion. There are exceptions. Constriction bands can restrict growth. Some syndactylies have bony fusion of the distal phalanx; if this is the long and ring fingers with a length discrepancy, the long finger can be markedly flexed and benefit from release during the first few months of life. The most powerful influence that will restrict growth is badly performed surgery. This can affect growth in a number of ways. If surgery reduces the use of the hand for any reason it will affect growth. Nerve damage reduces both growth and volume. As outlined above, badly placed scars and skin grafts cause contracture and effect growth. Worse still, dense scarring, bone or joint damage, or impaired vascularity may occur.

**Timing of surgery**

There are good arguments for surgery in the first year of life. It is the time when hand function, including prehension and coordination are developing. In a normal and otherwise healthy infant, not a small for dates infant, the tissues handle very well. In the first few months of life the skin is very stretchy, and the small amount of subcutaneous fat makes flaps and grafts more reliable. Nerve recovery is better the younger the child. In the very young the vessels seem disproportionally large, and microvascular anastomoses at this age are expected to be successful, although would usually be delayed if possible. The congenital hand surgeon will always operate with times 3.5 magnifying loops, so tissue size is not a problem.

This push for early surgery is not slavishly followed however as other factors come into play. The child may be premature or small for dates. It might have other medical or surgical conditions which take priority. Well planned hand surgery performed later than the generally considered optimal time should be successful.

Some operations need to be done in the first few months of life, for instance tight constrictions bands, floating polydactyly of the little finger or thumb, and some syndactylus cases with distant bony union and flexion deformities. The majority of congenital conditions are best having surgery between 6 and 18 months, for instance polydactyly, pollicisation of the index finger, syndactyly, and the longitudinal deficiencies such as radial club hands (subject to their haematology).

Some operations are long, for instance toe to hand transfers take five to ten hours, and are usually performed between 2 and 4 years of age. This allows time for the parents to make what is often a difficult decision, and for the anatomical size of the toe and vessels to become optimal.

In thumb aplasia there is debate as to whether a pollicisation performed at 6 months of age is substantially better than at 12 to 18 months or even later. There is no hard evidence either way but the quality of the surgery is probably the overriding factor.

Other conditions already mentioned in the context of therapy need surgery at a time when therapy, stretching, and splintage have achieved maximum effect.

Most conditions require long term follow up until fully grown. Approximately 5–20% of cases require secondary surgery. Common causes are scar tightness requiring Z-plasty lengthening or some other plastic procedure, joint instability for instance in hypoplastic or polydactyly thumbs, web space creep in syndactyly, minor adjustments to pollicisations, or toe to hand transfers such as tenolysis pulp plasty or scar revision.

New techniques give scope for further treatment, for example, the development of slow distraction techniques for positioning the hand in radial club hand.

**Notes on 11 congenital hand conditions**

**APERT’S MITTEN HAND**

The fourth web is released to give a little finger which is invariably virtually normal. Then the first web is released with full thickness grafts and skin flaps to give a good thumb, although often with a delta phalanx needing surgery in some cases. If the index, long, and ring fingers are good they may be separated, but often the bones are fused and the interphalangeal joints non-functional. In these cases it is best to leave the three central fingers joined together, giving the hand a tripod pinch (fig 1).

**ARTHROGRYPOSIS**

The hand surgeon must work closely with the orthopaedic surgeon looking after the child. The treatment follows all the broad aims outlined above. Surgery may have a place in some cases, although sadly is largely ineffective in the more severe cases. It is very important for
hand function to have at least one elbow that actively flexes, and muscle transfers may achieve this.

**Camptodactyly**
The anomaly is a flexed PIPJ. In its "pure" form the pipj is normal, it can be passively but not actively extended, and the metacarpal-phalangeal joint (MCPJ) is hyperextended. It is because of an absent or anomalous lumbrical muscle. Although small the lumbrical muscles are essential to balancing the MCP and pipj. The name covers a mixed bag of anomalies, some with abnormal PIPJs, with fixed flexion deformities. Therapy with night-time splintage in extension is the mainstay of treatment and often very effective. It has to be continued until maturity.

Some patients benefit from a tendon transfer, flexor digitorum sublimis of the little finger (FDS V) to the lumbrical insertion. Others benefit from release of the volar structures and pipj but the results of surgery can be unpredictable.

**Cleft Hand**
The typical cleft hand is usually bilateral, often affecting the feet as well, and the parts that remain are normal in size. In the most severe cases only a little finger remains. It is inherited as a dominant gene.

Atypical cleft hand is now considered as part of the spectrum of symbradyactyly. It varies in severity and the parts that remain are not normal in size. The deficiency is maximal on the ulnar side, leaving in severe cases only a hypoplastic thumb.

**Finger Hypoplasia and Aplasia**
Some length can be obtained by bone distraction techniques or by the controversial operation of free non-vascularised phalangeal transfer. The gain in length and function however is small. The best functional gain is from a vascularised toe to hand transfer, either single second toe or bilateral second toes\(^1\)\(^4\)\(^12\)\(^13\) (figs 2 and 3).

**Poland’s Anomaly**
The hand is affected to a varying degree and is treated accordingly. The absence of the sternocostal part of the pectoral muscle, and in females the breast, produces a very obvious cosmetic deformity. A cosmetic breast augmentation at maturity in the absence of any anterior chest wall muscle is not a very satisfactory procedure. Transfer of the latissimus dorsi muscle from the back to reconstruct the pectoralis major fills out the depression below the clavicle. It also allows a breast prosthesis to be placed beneath the muscle. It does produce a scar on the back and some contour loss, so this operation has to be considered in great detail by the patient and surgeon.

**Polydactyly**
Floating polydactylies are excised under local anaesthesia as a proper operation in the first few weeks. In most thumb polydactyly the radial thumb is smaller than the ulnar and the treatment is to excise the radial thumb. Where the two thumbs are a similar size the choice is to excise the radial, leaving a hypoplastic ulnar thumb which will inevitably have some Z angulation at the metacarpal-phalangeal joint (MCPJ) and interphalangeal joints (IPJ), or to amalgamate the two thumbs longitudinally in the Bilhaut–Cloquet operation.

**Radial Club Hand**
Therapy, stretching, and splintage must start in the neonatal period. The condition varies in severity from distal radial hypoplasia and a hand that responds well to therapy, to complete absence of the radius and a hand at right angles to the forearm, very tight, and responding poorly to therapy. The new techniques of slow distraction by external fixation devise are a genuine advance in the treatment. After slow distraction the hand can be aligned on the forearm by the traditional operations much more effectively. It is essential to preserve some wrist movement and to take great care of the distal ulnar epiphysis so as not to damage the forearm’s growth potential.

When the position of the hand has been improved to its maximum, the index finger is pollicised. Often the index finger in severe radial club hands is stiff and the results of pollicisation are less good.

**Ring Constrictions**
These are released by cutting large Z-plasties, which often have to be repeated as the finger grows.

**Syndactyly**
In the minority of cases the syndactyly is loose and can be released without full thickness skin grafts. This is especially true if the surgery takes place in the first few weeks of life when the skin is stretchy and there is little subcutaneous fat. Otherwise full thickness grafts are needed, interdigitated with finger flaps to minimise scar contraction. Pulp flaps provide a very effective reconstruction for the side of the nail.

**Thumb Hypoplasia and Aplasia**
Hypoplastic thumbs can be stabilised and given more movement by tendon transfers. In aplasia and severe hypoplasia there is no trapezium or trapezio-metacarpal joint, and a toe transfer is not appropriate. The operation of choice is to pollicise the index finger\(^5\)\(^6\) (fig 4).


### STAMPS IN PAEDIATRICS

Child welfare stamps

Some countries regularly issue child welfare stamps, often on an annual basis, with an incorporated surcharge to raise funds for a particular charity. The Argentine Republic issued regular child welfare stamps from 1958. Part of the 1970 set is inscribed “pro infancia”. New Zealand’s annual health stamp issues started in 1929 and frequently, although not invariably, featured children and raised funds for children’s health camps. “Children’s Health” has been specifically incorporated in the design in recent years. An example of an early health stamp from 1949 and one of the later issues from 1997 are shown.

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