Congenital hand anomalies

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Abstract

Congenital upper limb anomalies affect 0.1—0.2% of all newborns. They are often isolated phenomena but can be associated with other congenital anomalies and may be the only external manifestation of a syndrome. Knowledge of the treatment options is imperative to ensure appropriate referral and counselling.

The aim of surgery for a congenital hand anomaly is to improve both function and appearance. Apart from the face, the hand is the only other part of the body on regular display.

Independent living is largely dependent on good bimanual hand function. For example a large proportion of activities of daily living such as washing, dressing, and feeding consist of bimanual tasks. It is only when we temporarily lose the function of one hand that the significance of this becomes apparent. However, children with congenital hand anomalies adapt very well to limitations of hand function and can often find "trick" manoeuvres to achieve essential tasks.

As there is a wide variation in the types and severities of hand anomalies these cases are largely managed in specialized clinics. It is in this setting that the child and family will have access to long-term multidisciplinary care which includes input from geneticists, psychologists, therapists and children's hand surgeons. For some children with more complex anomalies, psychological support can be as valuable as surgery to aid integration with and acceptance by their peers.

Keywords birth defect; congenital hand anomalies; duplication; finger abnormalities; forearm abnormalities; hand deformity; newborn; syndactyly; thumb abnormalities; toe transfer

Treatment principles

Functionality and cosmesis

The aim of surgery or therapy is to provide the child with the best functional and cosmetic outcome possible. As some of these children have other anomalies it is critical to look at the global function of the child, rather than at specific elements of their hand function.

The best functional outcome will provide the child with normal grasp patterns for both single and bimanual tasks. In simple anomalies this can be achieved with relative ease. However in more complex and bilateral anomalies achieving an acceptable level of function for activities of daily living can be extremely challenging.

The limits of function are largely dependent on the number of competent digits in the hand and their location. The presence of a stable, sensate and mobile thumb is essential for competent hand function. The thumb plays a key role in achieving the varied grips possible — including key, pinch, large and small object grasp.

If the thumb is competent, then the focus can turn to the other digits.

In assessing and planning the reconstruction of a child's hand the surgeon must always plan for the future. A young child can function well with basic hand grasp patterns. However, as they grow and become more independent they will need more refined, precise hand movements.

Timing

There are very few congenital anomalies that require urgent surgical intervention although ring constriction syndrome with severe distal oedema and neonatal Volkmann's ischaemic contracture may be the exceptions.

However, referral to a specialized hand clinic where psychological, genetic and hand therapy input is available should be arranged as soon as possible after diagnosis.

Certain conditions merit very early surgery not necessarily because of the severity of the hand anomaly but more for the benefit of performing surgery under local anaesthesia in neonates. This is possible for some with extra digits and for the release of minor acrosyndactyly (the distal joining of finger tips in ring constriction syndrome).

Early surgery before the age of 1 year is recommended for the separation of syndactylized (joined) border digits including involvement of the first web space. This will prevent the problems related to differential growth of the digits and optimize hand function.

In general if surgery is performed before school age the child will tend to forget any negative psychological experience and make a speedy postoperative recovery.

However, if co-operation and compliance are essential for good postoperative outcome, as in tendon transfers, then it is best to wait unit the child is 6/7 years old.

Apart from these examples, surgery for the correction of most congenital anomalies takes place once the following criteria are satisfied: (i) each subsequent figure should be increased, the risk of general anaesthesia is minimal or as low as possible given any other organ anomalies; (ii) a knowledge of the severity of other anomalies is apparent and they have been treated where possible; (iii) the size of the hand structures is such that surgery is possible; (iv) sufficient time has elapsed so that the benefit of splinting has had time to be effective. i.e. clasp thumb, trigger thumb, camptodactyly.

In the majority of children this is between the ages of 1 and 2 years.

Psychology

The attitude of the child's parents affects their long-term outcome. Their approach and method of coping with their child's hand anomaly will significantly affect how the child adapts psychologically. This in turn will influence how the child integrates into society.

As the severity of the congenital abnormality may vary, so too can the reaction of the child's parents and does not necessarily parallel the severity of the child's hand anomaly.[1] Parents often have difficulty making decisions about surgery on behalf of their new baby. This can lead to parental disharmony and feelings of guilt.

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Early input from a psychologist with experience dealing with parents and children with hand anomalies can be very valuable in helping to allay fears and to encourage acceptance and support.[2,3]

Associated anomalies

Congenital hand anomalies can be part of a recognized genetic syndrome or they may occur as a consequence of arrest or disruption in gestational development. The limb bud develops between the 4th and 8th week of gestation. At this time other organs and systems are also developing so there can be disruption of both limb and other organs simultaneously. This explains why radial ray deficiencies can be associated with other anomalies of cardiac, skeletal and gastrointestinal systems as occurs in the VACTERL sequence (Vertebral, Anal, Cardiac, Tracheo-oesophageal, Renal and Limb).

Simultaneous proximal and distal disruptions of limb development may also occur as in Poland's syndrome where there can be associated anomalies of the hand with symbrachydactyly and complete or partial absence of the pectoral muscles.

Knowledge of the common syndromes and typically associated anomalies is very useful when assessing a child with a congenital hand anomaly. It can help in planning timing of surgery and ensuring appropriate specialist referral if other anomalies are diagnosed.

In some syndromes the limb anomaly may be the only external manifestation of the syndrome. This is the case in Thrombocytopenia Absent Radius syndrome and Fanconi 's Anaemia. The latter is a potentially fatal condition where the radial ray deficiency or duplication may be the only external feature of the syndrome. Children with Fanconi's Anaemia usually develop bone marrow failure within the first decade of life and are at high risk of solid organ malignancy. Early diagnosis of Fanconi's anaemia at the time of presentation of the limb anomaly is essential to ensure appropriate referral to haematology and genetic clinics for assessment and counselling.

Classification

In 1968 Swanson first classified congenital anomalies by their morphology assuming these were related to defects in embryogenesis.[4] This system has remained as the most commonly used classification system to date. It has been amended and updated with the progress of time and knowledge by Swanson, the International Federation of Societies for Surgery of the Hand (IFSSH) and the Japanese Society for Surgery of the Hand (JSSH).[5,6]

However there are still controversies about the classification of certain anomalies such as symbrachydactyly which was originally classified in group V as undergrowth while atypical cleft hand was classified in group I failure of formation. As the spectrum of congenital anomalies is so great there is, as one would expect, a group of unclassifiable anomalies which was added at the time of the JSSH modification to include all those anomalies which do not fit into another category.

As further knowledge about the embryological aetiology of various congenital hand anomalies unfolds a new classification system based on embrynogenesis may supersede this current system.

The Swanson classification system divides the anomalies into the following categories: Failure of formation, Failure of differentiation, Duplication, Overgrowth, Undergrowth, Constriction band syndrome and Generalized Skeletal Abnormalities.

Each category is then divided into subcategories for example group I failure of formation is divided into the subcategories: (Ia) longitudinal, (Ib) transverse or (Ic) failure of finger ray induction including cleft hand.

Each subcategory is then further catalogued by the anatomical level of the deformity i.e. shoulder, arm, elbow, wrist etc. Following this the anomalies are then listed by common diagnosis for example, cleft hand, cutaneous syndactyly (Table 1).

Failure of formation

Radial longitudinal deficiency: deficiencies on the radial border of the hand and arm are not common. They are significant not only because of the impairment of hand function but also because there is a high risk of associated anomalies. The risk of associated thumb hypoplasia is proportional to the severity of the radial deformity.[7] Radial border anomalies have a high incidence

Abbreviated IFFSH Swanson's classification of congenital hand anomalies

Main category	Subcategory	Diagnosis
I. Failure of formation	a. Transverse	
	b. Longitudinal	Longitudinal radial deficiency
		Longitudinal ulnar deficiency
	c. Central	Cleft hand
II. Failure of differentiation	a. Soft tissue	Cutaneous syndactyly
		Camptodactyly
		Congenital trigger digit
	b. Skeletal	Radioulnar synostosis
		Synostosis of the metacarpals
		Synostosis of the phalanges
		Symphalangia
		Clinodactyly
	c. Tumorous	Haemangioma
		Malformation
		Osteochondromatosis
		Enchrondroma
		Fibrous dysplasia
		Epiphyseal abnormalities
III. Duplication		Thumb duplication
		Ulnar polydactyly
IV. Overgrowth		Hemihypertrophy
		Macrodactyly
V. Undergrowth		Brachdactyly
		Brachysyndactyly
VI. Constriction Band Syndrome		Constriction band with oedema
		Constriction band without oedema
VII. Generalized abnormalities and syndromes		Marfan's syndrome

Table 1