



## **IFSSH Scientific Committee on Congenital Conditions**

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**Report presented November 2010, Seoul, Korea**

## **1: Congenital Limb Anomalies – Scott N. Oishi, Marybeth Ezaki**

### **1. The problem.**

The child born with a noticeable anomaly has been the focus of enormous natural curiosity throughout recorded history. These special children have been regarded as monsters in many societies, and as gods or goddesses in others. Cultural and religious views determine whether these children are accepted or reviled.

Whereas in the past these children were hidden away from the public, today children with special needs are included in schools, and supported with reasonable adaptations and focused therapy services mandated by the laws of many countries. Happily, today the birth of a child with a limb anomaly in the United States and developed nations is seen not as a curse, but as a condition that has a natural explanation. Most children with congenital upper limb anomalies have normal cognitive function, and therefore, have the potential to both understand and overcome their differences and to be productive members of our communities. Our society has come to accept these children, to recognize their needs, and to celebrate their successes.

Congenital anomalies of the upper limb present enormous challenges for the child, the parents, the family and the professionals caring for the child. In addition to the clinical findings that define and characterize the abnormal morphology of the upper limb, important aspects of evaluation and treatment must be directed to the identification of associated syndromes and anomalies, and the emotional and educational support of the parents and family. Later issues that must be addressed include all the adaptations that contribute to functional independence, an intact self-esteem, and psychological health for the child herself.

The problem that we, as hand surgeons, are faced with is how to best use our skills and knowledge to enhance the ability of any given patient to function in society. How can we know and do what is the best for the child.

### **2. The solution.**

Establishing an accurate and complete diagnosis is a team effort. Most of these small patients are treated at referral centers where expert care is available for the whole child. A network of dedicated pediatric orthopaedic hospitals was established by numerous benevolent organizations in the United States, initially to provide free care to the children crippled by poliomyelitis in the last century. Today, the Shriners Hospitals, the Scottish Rite Hospital for Children, and others with the same mission, continue to provide this care for children with orthopaedic needs. The upper limb surgeons from these hospitals have organized meetings for the collegial exchange of information as well as informal networking and referral of complex patients. Numerous publications have come from these collaborations.

The Hand Surgeon must be aware of the potential for associated anomalies and may be the one to initiate the appropriate referrals. With the hand surgeon should reside the

knowledge to know what to look for, especially as education in the primary care specialties devotes little or no time to most of these obscure conditions.

Help must also be made available to educate and support the parents in the critical bonding with their infant, especially if the diagnosis of the congenital anomaly was not made before birth. Divorce is a very common response to the birth of a child with a congenital limb anomaly because of the added stress on the marriage. The child herself will experience a normal grieving process when she is old enough to understand the full impact of having a limb anomaly. Family and caregiver support systems must be robust to nurture a child through these transition periods with a valued sense of self.

Care of the child with a congenital anomaly is complex and rewarding, and must be long term and ongoing. Enabling a child to interface with the environment and become more independent must be the goal of any treatment. The guiding principle of “To-For” in deciding whether an operation is indicated, recognizes that there are many things that we can do “To” the child, but we must make sure that what we do is truly “For” the child.

Amazing advances in surgical technique have added immensely to our ability to reconstruct and improve the lives of our patients. The judgment to know which child will benefit from microsurgical free toe transfers, or from the application of limb lengthening techniques is more difficult to learn. Where is the evidence? *Primum non nocere*

Children with congenital anomalies of all sorts will continue to be born as the result of genetic mutation, or genomic variation, epigenic effects, and teratogenic causes. Advances are being made, at an ever increasing rate, that define the basic science of the causes of congenital anomalies. The understanding of molecular mechanisms of causation may someday lead to the ability to prevent or alter final outcomes for children – but in the distant future. Not all of these anomalies will be predicted or prevented. We must continue to ask questions and ensure that our communities support research in the areas of congenital anomalies. We must, likewise, work toward ensuring a clean and healthy environment for the children of our children.

We must also recognize and fight the discrimination that comes from ignorance and ill-focused blame and guilt. Innocent curiosity that changes into schoolyard teasing, can easily change into deeply wounding harassment if we do not address current societal values about limb malformations. Because of our unique role in the lives of these children, we are able to ask the questions that will uncover these problems. “Does your hand ever make you sad?” “Who is your best friend?” “Draw a picture of yourself.” Early intervention with psychological support and peer groups is more critical than any surgical procedure we can do. Social networking is an important aspect of support for our patients. In the United States camps, retreats, websites, handouts, and peer support offer enormous benefits for the children and the families, with equal feedback and immense personal reward for the surgeon who chooses to become involved in these efforts.

As hand surgeons, we must perform a critical analysis of our own results and honestly report them so that we can move the science and care of the children forward. Congenital hand anomalies are unusual and our natural curiosity makes us eager to try a procedure that has appeared in the textbook. Long term follow-up and outcomes assessment of how we impact Quality of Life are much more important than short term measures of pinch and grip, and must become an integral part of what we do.

Many hand surgeons – too many to name - have contributed to the knowledge we have about congenital anomalies. National and international meetings provide opportunities to exchange information and updates. At these meetings, the name of the nation is “Science,” a land with no borders, and our anthem must be “For the Children.” Translational research that brings clinicians and basic scientists together will greatly expand our knowledge in this century. Rapid exchange of information and electronic consultation benefit us as well as our patients.

The future is bright for the field of congenital hand surgery. We must constantly remember to put the best interest of our patients first, and to follow and support them to the other side of their childhood.

(The manuscript above has been written by Dr. Marybeth Ezaki and Dr. Scott N. Oishi for the chapter for the IFSSH book, Hand Surgery –Worldwide: International Reconstruction of a “Beautiful and Ready Instrument of the Mind”. Dr. Urbaniak, who is a President of the IFSSH, allowed us to use it as our committee report.)

## **2: Upper Limb Hypertrophy**

The Committee discussed the appropriate diagnostic name for hyperplasia of the hand and upper limb with aberrant muscles. This condition is characterised by muscle hyperplasia, aberrant muscles or accessory muscles, ulnar drift of the fingers at the metacarpophalangeal joints, flexion contractures of the metacarpophalangeal joints, extension contractures of the wrist, and enlargement of the spaces between the metacarpals. The Committee proposes the descriptive term “aberrant muscle syndrome” or “accessory muscle syndrome” for this condition.

### **3: Triphalangeal thumb**

A small accessory middle phalanx in the skeletally immature may have a joint both proximal and distal to the accessory phalanx (type A). Alternatively there may be a cartilaginous connection between the accessory middle phalanx and the epiphysis of the terminal phalanx, which is not obvious radiologically in the skeletally immature (type B). Following completion of ossification, type A will proceed to a mature and separate middle phalanx (type D). However, type B will mature into a triangular epiphysis with no joint between the middle phalanx and terminal phalanx.

The Committee agrees that types B and C are part of the spectrum of thumb triphalangism.