Pediatric Limb Differences and Amputations

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KEYWORDS

• Pediatric limb difference • Congenital limb deficiency • Amputation

KEY POINTS

- Congenital limb differences are uncommon and often go undetected until birth.
- A thorough history, physical examination, or diagnostic workup should be done for children with congenital limb differences to rule out syndromes involving other organ systems or known associations.
- Acquired amputations most commonly occur from trauma.
- Complications, such as pain and terminal bony overgrowth can occur after amputation.
- A multidisciplinary approach to management is recommended, when available.

NATURE OF THE PROBLEM

The Centers for Disease Control and Prevention estimate that each year 2250 babies are born with congenital upper and/or lower limb deficiencies or reductions each year in the United States. This is approximately 6 per 10,000 live births per year, in a ratio of 2:1 upper to lower extremity.[1] Precise numbers for other forms of congenital limb differences (ie, limb length discrepancies, neuromuscular pathology leading to differences in limb) and joint deformities (ie, contractures) are not known. Recent data suggest a relationship between paternal occupation and increased prevalence of birth defects, including limb deficiencies, in offspring of artists.[2] No racial predilection has been noted. Medications known to affect limb development include thalidomide, retinoic acid, and misoprostol. Teratogenic causes are often challenging to discern, as prenatal history may be complicated by maternal recall bias, and the timing of limb development is coincident when the mother may not know she is pregnant.[3] Limb deficiencies can also be caused by vascular disruption (eg, amniotic band syndrome),

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vascular malformations (eg, Poland syndrome), or genetic factors (spontaneous point mutation). Findings from experimental animal studies suggest that limb deficiency in amniotic band syndrome may be caused by a cascade of hypoxia, cell damage, hemorrhage, tissue loss, and reperfusion.[4] In most cases the cause is unknown.[5]

Acquired amputations most commonly occur from trauma or disease (ie, neoplasm or infection.) A retrospective study done in the United States determined that there were more than 110,000 children younger than 18 years that presented to emergency rooms with traumatic amputation injuries during a 12-year period. The average age was 6.18 years, patients were predominantly males (65.5%), and finger amputations comprised 91.6% of the amputations.[6]

Despite prenatal screening ultrasound scans, congenital limb deficiencies may not be detected before birth. The International Organization for Standardization (ISO) names congenital limb deficiencies as follows: transverse (normal limb development to a particular level, with no skeletal elements distally) and longitudinal (absence or reduction of an element within the long axis).[7]

Terminology used to differentiate acquired amputations in the upper limb includes shoulder disarticulation, transhumeral, elbow disarticulation, transradial, wrist disarticulation, and partial hand amputation. The different forms of lower limb amputations are translumbar, transpelvic, hip disarticulation, transfemoral, knee disarticulation, transtibial, ankle disarticulation, and partial foot amputation.[8]

PATHOPHYSIOLOGY

Limb development occurs between 4 and 8 weeks after fertilization. Most limb defects are thought to occur during weeks 4 and 6, during times of rapid tissue proliferation.[9] Limb development is considered with respect to its 3 axes of growth (proximal-distal, anterior-posterior/radio-ulnar, and dorsal-ventral.) Each axis is controlled by distinct, yet coordinated, molecular pathways, which include fibroblast growth factors, sonic hedgehog, and the Wingless-type signaling pathways. Each pathway is responsible for its own differentiation, yet they work in concert and have complex interactions with signaling, regulation, feedback loops, and maintenance of the other axes and embryogenesis. Errors in these pathways can indirectly affect the appropriate operation of other signaling centers, which may reflect the presence of other organ systems involved in some children with limb deficiencies.[10]

CONGENITAL LIMB DIFFERENCES

Upper Limb

Polydactyly is a congenital hand difference resulting in an extra digit. The extra digit may be preaxial (radially located) postaxial (ulnarly located) or centrally located. Most cases of preaxial polydactyly are sporadic and occur unilaterally. However, if the extra thumb has 3 phalanges, it may be linked to a systemic syndrome, such as Holt-Oram syndrome or Fanconi anemia. Postaxial polydactyly is often found in patients of African or African-American descent and can be inherited in an autosomal dominant pattern. If the postaxial digit is found in a Caucasian patient, there may be an underlying syndrome, such as chondroectodermal dysplasia or Ellis-van Creveld syndrome.

Syndactyly is a condition in which the digits fail to separate into individual appendages. Syndactyly can be simple, in which only the soft tissues are involved, or it can be complex, in which the bone or nail of the neighboring fingers is involved. It is typically an isolated finding; however, it may be associated with certain syndromes, such as Apert or Poland syndrome. Apert syndrome, or acrocephalosyndactyly, is characterized by