Arthrogryposis

Objectives
1. Define arthrogryposis multiplex congenita
2. Describe clinical features of arthrogryposis multiplex congenita
3. Discuss genetic transmission and pathology of arthrogryposis multiplex congenita
4. Discuss management of the upper limb, hip, knee, and foot problems associated with arthrogryposis multiplex congenita

Discussion

Arthrogryposis multiplex congenita is a confusing entity, probably still heterogeneous as syndromes producing a similar clinical picture are identified; and subgroups, such as distal arthrogryposis are better defined. Goldberg listed criteria he felt necessary to define arthrogryposis multiplex congenita: 1) a characteristic tubular limb posture with absent flexion creases, adducted and internally rotated shoulders, (usually) extended elbows, flexed and ulnarly deviated wrists, tapered and flexed fingers, hyperflexed hips, knees fixed in flexion or hyperextension, and clubfeet 2) typical face, which is not dysmorphic 3) normal intelligence and an absence of visceral abnormalities, and 4) a negative family history. Staheli and colleagues at the University of Washington term this type of arthrogryposis amyoplasia. To confuse the issue, a number of reports with phenotypes appearing to be arthrogryposis are published with varying genetic patterns, whether or not these are all the same or related entities is unsettled at present. A particularly well defined subtype of arthrogryposis is distal arthrogryposis. Pathologic reports document abnormalities in the anterior horn cells, while in spinal muscular atrophy, the anterior horn cells were diminished or absent; they were increased and disorganized in arthrogryposis.

Management of children with arthrogryposis can be trying, but rewarding. Most, but not all, will become ambulatory. For the clubfoot, standard clubfoot surgical release, talactomy, and subchondral resection of the talus and cuboid have their advocates. Both good and poor results have been reported after open reduction of fixed hip dislocations. Upper limb surgery also has mixed results.

In summary, there is a distinct entity recognized by orthopaedic surgeons as arthrogryposis multiplex congenita. The exact genotype of this disorder is not established at present, most cases appear to be spontaneous; yet genetic patterns of transmission of arthrogryposis like phenotypes are regularly reported. The basic pathology appears to be in the anterior horn cells. The combination of contracture,
dislocation, and weakness represent difficult challenges to treatment, but the intelligence and perseverance of the patient population result in a high level of function despite disability. The rather extensive list of references reflects the varied approaches to the study and management of arthrogryposis.

References


