Summary

Arthrogryposis, or arthrogryposis multiplex congenita, comprises nonprogressive conditions characterized by multiple joint contractures found throughout the body at birth (A contracture is a limitation in the range of motion of a joint.). Children with the disorder are stiff and have many joint contractures. Creases in the skin are often lost; instead, small dimples can be seen in the skin over the joints.

Arthrogryposis can occur for many reasons. One reason is a primary neuropathic defect. However, in all cases of arthrogryposis, for one reason or another, fetal movement has decreased. Motion is essential for normal development of joints and their adjoining structures. Lack of movement during development of the embryo causes extra connective tissue to develop around the joint. This results in fixation of the joint, limiting movement and further aggravating joint contracture.

Diagnosis is made by ruling out other causes. Muscle biopsies, blood tests, and clinical findings help rule out other possible disorders and provide evidence for arthrogryposis. There is a wide variation in the degree to which muscles and joints are affected in those with arthrogryposis.

The skin of children with arthrogryposis is thin and smooth. The underlying muscle is pale and thin, and often replaced by fat or fibrous tissue. The capsules and legaments of the joints are thickened and the articular surfaces are abnormally shaped. Narrowing of the spinal cord in the cervical and lumber regions with increase in the ventricular size has been described.

The combination of contracture, dislocation, and weakness represent difficult challenges to treatment.

Treatment of arthrogryposis is often challenging and should not be undertaken by any one doctor, but by a multi-disciplinary team, including the pediatrician, neurologist, orthopedic surgeon, geneticist, physical and occupational therapist. The aim of treatment is to improve function. Intervention includes therapy, splinting, and surgical procedures. To date there is no completely successful treatment approach to arthrogryposis. However, goals for intervention include lower limb alignment and establishment for ambulation and upper limb function for self-care. Furthermore, with therapy and other available treatments, substantial improvement in function is usually possible. Most people with arthrogryposis are of normal intelligence and are able to lead productive, independent lives as adults.