Pediatric Neurology Part III: Chapter 138. Arthrogryposis and fetal hypomobility syndrome (Handbook of Clinical Neurology)

Goknur Haliloglu, Haluk Topaloglu

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Arthrogryposis is a heterogeneous condition, evident from birth, which can be defined as multiple contractures of the joints. The etiology is multifold: genetic disorders of the central or peripheral nervous system, or of the connective tissue leading to decreased fetal movements, and vascular and environmental causes. The problem begins in utero. There may be overlapping conditions between sporadic, syndromic, neurogenic, myopathic and metabolic types. The workup should include a family tree. Systemic involvement, for example of the renal and pulmonary systems, may be encountered in associated syndromes. Motor neuron disorders leading to the condition are the most commonly seen type. Fetal or neonatal akinesia/hypokinesia is at the severe end of the spectrum, in which there is literally intrauterine limitation of movement. Children with amyplasia are born with little or diminished muscle bulk of the extremities. Distal arthrogryposis is almost always a dominantly inherited condition. A multidisciplinary care approach is required in order to provide optimum healthcare. The management team should include a nutritionist and a physiotherapist. Genetic counseling is possible in most instances. A truly genetic cause can be identified in more than 50% of cases. Survivors, though handicapped, can lead near normal lives.

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