

Arthrogryposis is a general or descriptive term for the development of nonprogressive contractures affecting one or more areas of the body prior to birth (congenitally). A contracture is a condition in which a joint becomes permanently fixed in a bent (flexed) or straightened (extended) position, completely or partially restricting the movement of the affected joint. When congenital contractures occur only in one body area, it is not referred to as arthrogryposis but rather an isolated congenital contracture. The most common form of an isolated congenital contracture is clubfoot. When arthrogryposis affects two or more different areas of the body, it may be referred to as arthrogryposis multiplex congenita (AMC). The most common form of AMC is amyoplasia. Arthrogryposis and arthrogryposis multiplex congenita are sometimes used interchangeably. The number of males and females affected by AMC is approximately equal. The condition has been reported in individuals of Asian, African and European descent. Isolated congenital contractures affect approximately 1 in 500 individuals in the general population. AMC affects approximately 1 in 3,000 individuals. AMC is, by definition, present at birth (congenital). A diagnosis of AMC is made based upon identification of characteristic symptoms (e.g., multiple congenital contractures), a detailed patient history, and a thorough clinical evaluation. Certain tests may be necessary to determine the underlying cause of AMC including nerve conduction, electromyography and muscle biopsy, which can help diagnose neuropathic or myopathic disorders. A nerve conduction study measures how rapidly nerves carry an electrical impulse. An electromyography is a test that records electrical activity in skeletal voluntary muscles at rest and during muscle contraction. A biopsy is a procedure in which a small amount of affected tissue (e.g., muscle) is removed and studied under a microscope to detect characteristic changes or findings that can aid in obtaining a diagnosis. Imaging studies of the central nervous system (CNS) and comparative genomic hybridization (CGH) array, microarray, and exome studies may also be useful studies in making diagnoses. Because of the many mutations that can lead to arthrogryposis, whole genome sequencing is often required (preferable in both parents for comparison as well) to make a diagnosis.