Natural history study of Arthrogryposis Multiplex Congenita, Amyoplasia type

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Arthrogryposis or Arthrogryposis Multiplex Congenita (AMC) are terms used to describe the clinical finding of multiple congenital contractures. There are more than 300 distinct disorders associated with arthrogryposis. Amyoplasia is the most common type of arthrogryposis and is often referred to as the “classic” type. There is no known cause of amyoplasia and no risk factors have been identified. Additionally, there is no established diagnostic criteria, which has led to inconsistency and confusion in the medical literature. The purpose of this study was to describe the natural history of amyoplasia, determine if there are any identifiable risk factors and develop a list of diagnostic criteria. A retrospective chart review of 59 children with arthrogryposis ascertained at the Shriners Hospitals for Children in Houston, Texas was performed and included the following information: prenatal, birth, and family histories, and phenotypic descriptions. Forty-four children were identified with amyoplasia and 15 children with other multiple congenital contractures and other anomalies (MCC) were used as a comparison group. With the exception of abnormal amniotic fluid levels during pregnancy, there were no significant demographic or prenatal risk factors identified. We found common features that discriminate amyoplasia from other types of arthrogryposis and developed a diagnostic checklist. This combination of the clinical features should now be used as diagnostic criteria for amyoplasia.

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