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Amyoplasia revisited

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Abstract

Amyoplasia is a specific type and the most common form of arthrogryposis (multiple congenital contractures). It is a clinical diagnosis at this time. Care should be used making the diagnosis because of the implications for recurrence, natural history, associated anomalies, and both etiology and pathogenesis. We reviewed over 600 published reports and 2,500 individual records to identify the 560 individuals reported here. Affected limbs had characteristic positions with fatty-fibrous replacement of muscle. Upper limb involvement was usually characterized by extended elbows. Lower limbs were held in various positions at birth; however, equinovarus positioning of feet was almost always present. Symmetric involvement was common. Among 560 affected individuals, subtypes were identified: four-limb symmetric involvement (331/560 = 55.9%), severe involvement (41/560 = 7.3%), three-limb involvement (27/560 = 4.8%), upper limb only Amyoplasia (ULA; 94/560 = 16.8%), and lower limb only Amyoplasia (LLA; 25/560 = 15.5%). Discordant monozygotic twinning was increased, occurring in 6.6% (37/560; OR 10.9). A variety of additional anomalies were seen, attributed to apparent vascular compromise. Gastrointestinal vascular compromise-type anomalies were present in 9.1% (51/560), trunk muscle defects in another 2.7% (15/560), digit compromise in 12.1% (68/560), constriction rings in 4.3% (24/560), and perinatal long bone fractures in 10.5% (59/560). Although prenatal ultrasound became the standard of care in 1990, only about one quarter of affected pregnancies were diagnosed prenatally since 1990. Amyoplasia appears to be completely sporadic. Novel pathogenetic mechanisms for the congenital anomalies seen in Amyoplasia need to be identified.

Keywords: Amyoplasia; arms only; arthrogryposis; bowel atresia; club hands; clubfeet; digit loss; dislocated hips; gastroschisis; hemangioma; hyperextension; legs only; monozygotic twins; multiple congenital contractures; pregnancy complication; prenatal diagnosis; twins; vascular compromise.

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