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The Core Curriculum

Arthrogryposis

Objectives

1. Define arthrogryposis multiplex congenita
2. Describe clinical features of arthrogryposis multiplex congenita
3. Discuss genetic transmission and pathology of arthrogryposis multiplex congenita
4. Discuss management of the upper limb, hip, knee, and foot problems associated with arthrogryposis multiplex congenita

Discussion

Arthrogryposis multiplex congenita is a confusing entity, probably still heterogenous as syndromes producing a similar clinical picture are identified; and subgroups, such as distal arthrogryposis are better defined. Goldberg listed criteria he felt necessary to define arthrogryposis multiplex congenita: 1) a characteristic tubular limb posture with absent flexion creases, adducted and internally rotated shoulders, (usually) extended elbows, flexed and ulnarly deviated wrists, tapered and flexed fingers, hyperflexed hips, knees fixed in flexion or hyperextension, and clubfeet 2) typical face, which is not dysmorphic 3) normal intelligence and an absence of visceral abnormalities, and 4) a negative family history. Staheli and colleagues at the University of Washington term this type of arthrogryposis amyoplasia. To confuse the issue, a number of reports with phenotypes appearing to be arthrogryposis are published with varying genetic patterns, whether or not these are all the same or related entities is unsettled at present. A particularly well defined subtype of arthrogryposis is distal arthrogryposis. Pathologic reports document abnormalities in the anterior horn cells, while in spinal muscular atrophy, the anterior horn cells were diminished or absent; they were increased and disorganized in arthrogryposis.

Management of children with arthrogryposis can be trying, but rewarding. Most, but not all, will become ambulatory. For the clubfoot, standard clubfoot surgical release, talectomy, and subchondral resection of the talus and cuboid have their advocates. Both good and poor results have been reported after open reduction of fixed hip dislocations. Upper limb surgery also has mixed results.

In summary, there is a distinct entity recognized by orthopaedic surgeons as arthrogryposis multiplex congenita. The exact genotype of this disorder is not established at present, most cases appear to be spontaneous; yet genetic patterns of transmission of arthrogryposis like phenotypes are regularly reported. The basic pathology appears to be in the anterior horn cells. The combination of contracture,

dislocation, and weakness represent difficult challenges to treatment, but the intelligence and perseverance of the patient population result in a high level of function despite disability. The rather extensive list of references reflects the varied approaches to the study and management of arthrogryposis.

References

1. Akazawa H, Oda K, Mitani S, Yoshitaka T, Asaumi K, Inoue H. Surgical management of hip dislocation in children with arthrogryposis multiplex congenita. *Journal of Bone & Joint Surgery - British Volume* 1998;80(4):636-40.
2. Bamshad M, Jorde LB, Carey JC. A revised and extended classification of the distal arthrogryposes. *American Journal of Medical Genetics* 1996;65(4):277-81.
3. Banker BQ. Arthrogryposis multiplex congenita: spectrum of pathologic changes. *Human Pathology* 1986;17(7):656-72.
4. Clarren SK, Hall JG. Neuropathologic findings in the spinal cords of 10 infants with arthrogryposis. *Journal of the Neurological Sciences* 1983;58(1):89-102.
5. D'Souza H, Aroojis A, Chawara GS. Talectomy in arthrogryposis: analysis of results. *Journal of Pediatric Orthopedics* 1998;18(6):760-4.
6. Daher YH, Lonstein JE, Winter RB, Moe JH. Spinal deformities in patients with arthrogryposis. A review of 16 patients. *Spine* 1985;10(7):609-13.
7. DelBello DA, Watts HG. Distal femoral extension osteotomy for knee flexion contracture in patients with arthrogryposis. *Journal of Pediatric Orthopedics* 1996;16(1):122-6.
8. Gruel CR, Birch JG, Roach JW, Herring JA. Teratologic dislocation of the hip. *Journal of Pediatric Orthopedics* 1986;6(6):693-702.
9. Mennen U. Early corrective surgery of the wrist and elbow in arthrogryposis multiplex congenita. *Journal of Hand Surgery - British Volume* 1993;18(3):304-7.
10. Niki H, Staheli LT, Mosca VS. Management of clubfoot deformity in amyoplasia. *Journal of Pediatric Orthopedics* 1997;17(6):803-7.
11. Sarralde A, Reynoso MC, Nazara Z, Soto F, Hernandez A. Prenatal growth retardation, pelvic hypoplasia, and arthrogryptic changes of lower limbs: a distinct autosomal-recessive disorder. *American Journal of Medical Genetics* 1998;75(5):453-60.
12. Sarwark JF, MacEwen GD, Scott CI, Jr. Amyoplasia (a common form of arthrogryposis). *Journal of Bone & Joint Surgery - American Volume* 1990;72(3):465-9.
13. Sells JM, Jaffe KM, Hall JG. Amyoplasia, the most common type of arthrogryposis: the potential for good outcome. *Pediatrics* 1996;97(2):225-31.
14. Shohat M, Lotan R, Magal N, Shohat T, Fischel-Ghodsian N, Rotter JI, et al. A gene for arthrogryposis multiplex congenita neuropathic type is linked to D5S394 on chromosome 5qter. *American Journal of Human Genetics* 1997;61(5):1139-43.

15. Sodergard J, Ryoppy S. The knee in arthrogryposis multiplex congenita. *Journal of Pediatric Orthopedics* 1990;10(2):177-82.
16. Sodergard J, Ryoppy S. Foot deformities in arthrogryposis multiplex congenita. *Journal of Pediatric Orthopedics* 1994;14(6):768-72.
17. Solund K, Sonne-Holm S, Kjolbye JE. Talectomy for equinovarus deformity in arthrogryposis. A 13 (2-20) year review of 17 feet. *Acta Orthopaedica Scandinavica* 1991;62(4):372-4.
18. Spires TD, Gross RH, Low W, Barringer W. Management of the resistant myelodysplastic or arthrogryptic clubfoot with the Verebelyi-Ogston procedure. *Journal of Pediatric Orthopedics* 1984;4(6):705-10.
19. Staheli LT, Chew DE, Elliott JS, Mosca VS. Management of hip dislocations in children with arthrogryposis. *Journal of Pediatric Orthopedics* 1987;7(6):681-5.
20. Staheli LT. *Arthrogryposis- a text atlas*. New York: Cambridge University Press; 1998.
21. Szoke G, Staheli LT, Jaffe K, Hall JG. Medial-approach open reduction of hip dislocation in amyoplasia-type arthrogryposis. *Journal of Pediatric Orthopedics* 1996;16(1):127-30.
22. Thompson GH, Bilenker RM. Comprehensive management of arthrogryposis multiplex congenita. *Clinical Orthopaedics & Related Research* 1985(194):6-14.
23. Van Heest A, Waters PM, Simmons BP. Surgical treatment of arthrogryposis of the elbow. *Journal of Hand Surgery - American Volume* 1998;23(6):1063-70.
24. Zori RT, Gardner JL, Zhang J, Mullan MJ, Shah R, Osborn AR, et al. Newly described form of X-linked arthrogryposis maps to the long arm of the human X chromosome. *American Journal of Medical Genetics* 1998;78(5):450-4.