Notes on spinal and scapular surgery

Excerpted from Muscular Dystrophy: A Concise Guide. Ed. Raymond A. Huml, Springer 2015.

Infantile Facioscapulohumeral Muscular Dystrophy - R.K. Lark and E.W. Hubbard

Infantile facioscapulohumeral muscular dystrophy (iFSHD) has become increasingly recognized as a related but distinct disease process over the past several decades [31 , 32 , 56 , 60 – 63]. Although molecular evaluation has shown that the same gene is affected as in adolescent facioscapulohumeral dystrophy, the disease course is more rapid and clinically this mirrors more severe MDs, such as Duchenne [61]. Infants usually develop facial diplegia [62]. Children begin walking at a normal age, but they rapidly develop pelvic girdle weakness. Children can have a positive Gower's sign and demonstrate significant hip extensor weakness on exam. They develop marked, severe hyperlordosis and use their hands to help stabilize their hip extensors while standing and walking, which is a near pathognomonic sign of this disease [63]. An equinus foot position develops, usually as a compensatory measure for quadriceps and tibialis anterior weakness. Patients develop shoulder girdle weakness, but unlike their adolescent and adult counterparts, it is the pelvic girdle weakness that dictates treatment in these patients.

The overall treatment goal for patients with iFSHD is to preserve function. Although hyperlordosis is severe, these patients do not respond well to bracing and use of orthoses should be limited. The hyperlordosis is a compensatory development, meant to counter the severe hip extensor weakness in these patients. Correcting the hyperlordosis with a brace or with surgery can actually inhibit ambulation because the patient can no longer compensate for their pelvic girdle weakness [33 , 63]. Similarly, the equinus foot position is also a compensatory measure. Most children respond well to ankle-foot orthoses or knee-ankle-foot orthoses, but on the rare occasion when a patient develops a rigid equinovarus foot position, an intramuscular lengthening could be considered [33 , 63].

Most patients lose the ability to ambulate by the second or third decade [60 – 62]. If the hyperlordosis is severe at that point, one could consider either bracing or surgical intervention to improve the patient's ability to sit in a wheel chair [33]. The shoulder girdle weakness is typically not a limiting condition for these patients because their spine and lower extremity conditions tend to be more severe. If the shoulder girdle weakness does interfere significantly with daily function, scapular stabilization can be considered. Scapulopexy is favored over scapulorthoracic arthrodesis in these settings because it allows immediate shoulder range of motion, there is no post-operative immobilization, and the procedure has less of an effect on pulmonary function, which is significantly more limited in patients with infantile facioscapulohumeral dystrophy. Scapular stabilization is not routinely performed in these patients because their disease course is so rapid, and the patients lose function in their upper extremities so quickly that the surgical benefits would not outweigh surgical risks to make the procedure worthwhile [32].

31. Lovell WW, Weinstein SL, Flynn JM. Lovell and Winter's pediatric orthopaedics. 7th ed.Philadelphia: Wolters Kluwer Health/Lippincott Williams & Wilkins; 2014.
32. Herring JA, Tachdjian MO. Texas Scottish Rite Hospital for Children. Tachdjian's pediatric orthopaedics. 4th ed. Philadelphia: Saunders/Elsevier; 2008.
33. Shapiro F, Specht L. The diagnosis and orthopaedic treatment of inherited muscular diseases of childhood. J Bone Joint Surg Am. 1993;75(3):439–54.
56. Birch JG. Orthopedic management of neuromuscular disorders in children. Semin Pediatr Neurol. 1998;5(2):78–91.
60. Bailey RO, Marzulo DC, Hans MB. Infantile facioscapulohumeral muscular dystrophy: new observations. Acta Neurol Scand. 1986;74(1):51–8.
61. Brouwer OF, Padberg GW, Wijmenga C, Frants RR. Facioscapulohumeral muscular dystrophy in early childhood. Arch Neurol. 1994;51(4):387–94.
62. Korf BR, Bresnan MJ, Shapiro F, Sotrel A, Abroms IF. Facioscapulohumeral dystrophy presenting in infancy with facial diplegia and sensorineural deafness. Ann Neurol. 1985;17(5): 513–6.
63. Shapiro F, Specht L, Korf BR. Locomotor problems in infantile facioscapulohumeral muscular dystrophy. Retrospective study of 9 patients. Acta Orthop Scand. 1991;62(4):367–71.