

Locomotor problems in infantile facioscapulohumeral muscular dystrophy

Retrospective study of 9 patients

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A retrospective study of 9 patients with infantile facioscapulohumeral muscular dystrophy defines orthopedic deformities and progression. Patients presented in the early months of life with facial diplegia. Sensorineural hearing loss occurred in 8 out of 9 with a mean onset at 5 (2-9) years. Walking began at the normal time, but worsened progressively, which was due mainly to gluteus

maximus muscle weakness. Scapular winging, extreme lumbar lordosis, and foot drop were characteristic. The majority of patients (in this and other series) lose walking ability in the second decade. Efforts to control lumbar lordosis by bracing while the patients were still walking were ineffective. Control of lumbar lordosis after the patients are wheelchair-dependent is important.

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Infantile facioscapulohumeral muscular dystrophy (IFSH MD) is characterized by (1) facial diplegia; (2) muscle weakness leading to winging of the scapulae, marked lumbar lordosis, and foot drop; and (3) sensorineural hearing loss (Carroll and Brooke 1979, Korf et al. 1985, Bailey et al. 1986). Occasionally, there is visual impairment due to retinal telangiectasia (Coats' disease; Small 1968, Taylor et al. 1982, Wulff et al. 1982). The prognosis in IFSH MD is much worse than in the more common facioscapulohumeral muscular dystrophy (FSH MD), where a relatively benign course leads to full life expectancy and maintenance of independent walking into late adult life. Neurologic aspects in 6 patients with IFSH MD from our unit have been reported by Korf et al. (1985).

In this article, we describe the orthopedic deformities and their consequences updating the clinical course in those patients previously reported and in 3 additional patients.

Patients and methods

Nine patients with IFSH MD followed at our Neuromuscular Diseases Clinic are reported. Factors assessed included sex, facial weakness, hearing deficits, age at initial walking, age at cessation of walking, scapular winging, lumbar lordosis, scoliosis, hip flexion contractures, foot deformities, and forced vital capacity expressed as a percentage of normal to indicate pulmonary function (Table 1).

Results

In our 9 cases, 6 became totally nonambulatory between 12 and 19 years of age. One 23-year-old man is still able to walk, but with increasing difficulty, spending much of his time in his wheelchair. The other 2 ambulatory patients are 7 and 10 years of age, but are having progressive difficulties. Although severe and eventually fixed lordosis invariably occurs, there is little problem with scoliosis. Six patients demonstrated no scoliosis by clinical examination. In 2 patients who had some deformity, radiography documented only a 14° T12-L3 deformity at 9 years of age in Case 9 and a 40° T8-L4 curve at 22 years of age in Case 3. In each of the 6 patients who had pulmonary function studies performed, there were decreases in forced vital capacity, with the values ranging from 12 to 68 percent of normal. Eight patients had sensorineural hearing deficits, and all of them benefited from hearing aids. The hearing deficit was diagnosed at a mean age of 5 (2-9) years. Case 3 has retained normal hearing at 22 years of age. No patient had retinal telangiectasia.

Case 3 has a pectus excavatum successfully repaired at 12 years of age. She also had a right scapular stabilization procedure at 14 years of age, but with little success. Case 5 (Figure 1) had a right tibialis posterior tendon transfer at 13 years of age, but progressive weakness led to recurrence of the deformity within 2 years. Since becoming wheelchair-bound, he has used a well-padded spinal orthosis to minimize his lumbar lordosis and to allow more

Table 1. Observations in 9 patients with infantile facioscapulohumeral muscular dystrophy

A	B	C	D	E	F	G	H	I	J	K	L	M	N
1	F	1	2	12	15		15	+	2		1	1	
2	F	3	7	14	12		14	+	3	90	1	0	68 (14)
3	F	24	-	12	19		22	+	3	10	0	2 ^a	62 (22)
4	F	0	5	21		7	7	+	2	0	1	0	
5	M	5	4	13	14		21	+	3	50	3	0	18 (20)
6	M	24	9	14		23	23	+	3	40	1	0	26 (23)
7	F	12	3	11		10	10	+	2	25	1	0	
8	F	5	5	12	13		19	+	3		1	0	66 (13)
9	F	3	8	18	12		14	+	3	90	1	1 ^b	12 (14)

A Case

B Sex

C Age facial weakness noted, months

D Age at diagnosis of sensorineural hearing loss, years.
- normal

E Age patient walked, months

F Age walking ceased, years

G Age still walking, years

H Age at latest assessment, years

I Scapular winging

J Lumbar lordosis: 1 +, 2 ++, 3 +++

K Hip flexion contracture, degrees

L Dorsiflexion-eversion weakness: 0 normal, 1 +, 2 ++, 3 +++

M Clinical scoliosis: 0 none, 1 +, 2 ++ 3 +++
^a40° left T8 to L4 curve at 22 years
^b14° right T12 to L3 curve at 9 years

N Forced vital capacity as percentage of normal (age in years)

comfortable seating. Case 6 had heel-cord and hamstring lengthenings respectively 12 and 15 years ago and remains ambulatory at 23 years of age. Two patients (Cases 8 and 9) had spinal bracing attempted while they were still able to walk, but neither one of them was able to tolerate the brace, walked less effectively with it on, and soon discontinued using it.

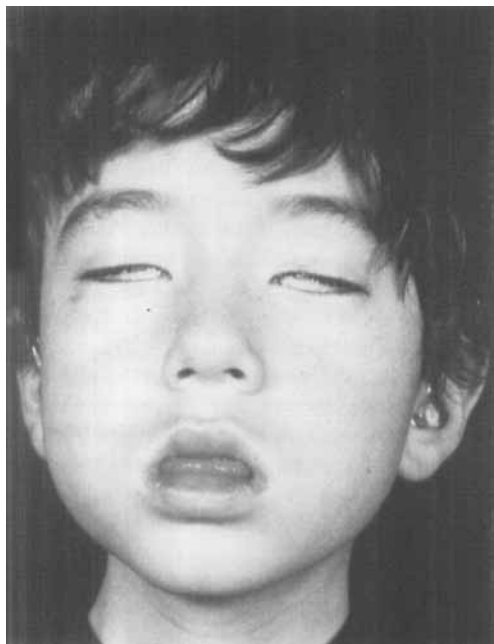
Discussion

Early onset of IFSH MD has been recognized for over a century. Landouzy and Dejerine (1884) described the childhood form presenting with facial weakness, and Duchenne commented on the different pattern of some cases compared with more commonly seen muscle disease variants (Carroll and Brooke 1979, Bailey et al. 1986). Hanson and Rowland (1971) pointed out the differentiation from conditions classified as part of the Möbius syndrome, in which the bifacial weakness alone predominates. It is has only been during the past few years, however, that IFSH MD has been clearly recognized as a distinct and much more severe condition than the more commonly occurring FSH MD; the latter is generally not diagnosed before 7 years of age, and in most instances is discovered on the basis of clinical criteria in the second decade of life, with some afflicted persons showing minimal symptoms throughout adult life (Tyler and Stephens 1950, Kazakov et al. 1974). The large majority of FSH MD patients have a normal life expectancy, remain

ambulatory, and do not have hearing or visual defects. The most common orthopedic intervention in FSH MD is scapular stabilization (Ketenjian 1978).

IFSH MD presents in infancy with bifacial weakness, a weak suck reflex, gagging, an expressionless face, and, later on, an inability to close the eyes, smile, whistle, or speak clearly. Six of 11 patients in the Carroll and Brooke (1979) group also had sensorineural deafness, as had most of our patients. None of our patients had visual difficulties due to retinal telangiectasia as described by Small et al. (1968), Taylor et al. (1982), Wulff et al. (1982), Fitzsimons et al. (1987), and Yasukohchi et al. (1988).

In IFSH MD, walking generally begins within the normal time frame; 7 of our 9 patients walked between 11 and 14 months. In the Carroll and Brooke (1979) series, 8 of 11 walked between 9 and 15 months of age, whereas the others walked at 16, 19, and 22 months. Once walking has begun, however, the children progressively worsen in comparison with their normal age-mates. The most significant limitation to walking is progressive weakness of the gluteus maximus and medius muscles in the presence of stronger hip flexors and progressive weakness of the anterior abdominal musculature in the presence of slightly stronger spinal extensor muscles. This results in the most severe lumbar lordosis, encountered in the entire spectrum of neuromuscular disorders. As illustrated in Figure 1, patients invariably stand and walk, towards the end of the first decade, with the hands and forearms folded across the upper buttocks to provide support for the failing gluteus maximus. This



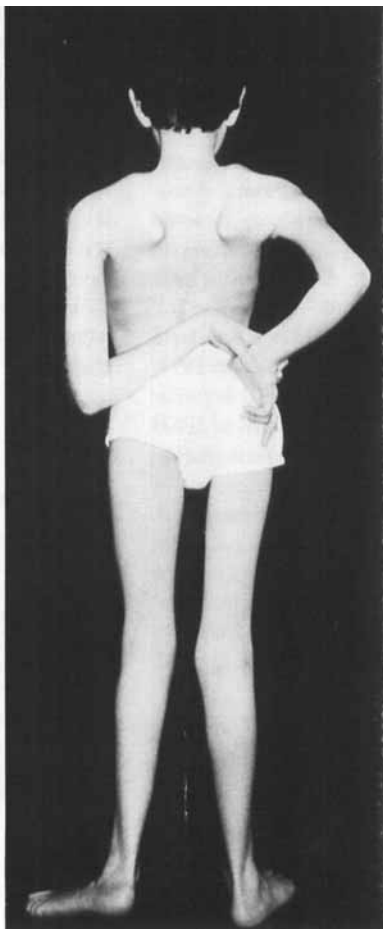
gait pattern is virtually pathognomonic for IFSH MD in our experience. The lumbar lordosis remains flexible for a few years, but it usually becomes rigid, especially after wheelchair dependency. Once the flexion deformities of the hip become rigid, contractures often reach 90°. Weakness of foot dorsiflexion and eversion is almost always seen, but rigid

Figure 1. Case 5, a 13-year-old boy with infantile facioscapulohumeral muscular dystrophy.

Facial photograph showing bifacial weakness. Note the inability to completely close the eyes and mouth. A hearing aid is visible in the left ear.

Lateral photograph with the patient standing shows the extremely marked lumbar lordosis with the hands clasped on posterior hip region for support. This clinical appearance is virtually pathognomonic for infantile facioscapulohumeral muscular dystrophy.

Back view of patient showing the prominent scapular winging, lordosis, and use of forearms and hands for support.



equinovarus deformities have developed only in 1 patient (Case 5). The large majority of the patients become nonambulatory in the second decade of life. Although severe and eventually fixed lordosis is an invariable component of the IFSH MD condition, there is little problem with scoliosis.

Abdominal, thoracic, and spinal extensor muscle weakness seriously compromise respiratory function. Death due to pulmonary failure has been reported in a 5-year-old child with IFSH MD (McGarry et al. 1983), in 4 affected family members each of whom died during the teenage years (Bailey et al. 1986), and in a 13-year-old (Yasukohchi et al. 1988).

Neuromuscular workup in IFSH MD reveals a myopathic EMG, mildly to moderately elevated CPK and aldolase levels, and a myopathic histologic appearance in biopsy specimens (Carroll and Brooke 1979, Korf et al. 1985).

Classical FSH MD is inherited as an autosomal dominant trait (Tyler and Stephens 1950). The inheritance of IFSH MD is less clear. Three parents of patients in our study had evidence of mild involvement, in one instance detected only as a subtle EMG abnormality in the deltoid (Korf et al. 1985). Eight of 11 patients reported by Carroll and Brooke (1979) had a parent with evidence of mild FSH MD. Gurwin et al. (1985) described a sibship of 3 with IFSH MD, Coats' disease, and hearing loss in which the mother had mild muscle weakness and tortuosity of the retinal vessels. These findings are compatible with autosomal dominant transmission with variable expressivity in some families. On the other hand, IFSH MD occurring in sibships without apparent muscle disease in the parents has been reported by Small (1968) and by Yasukohchi et al. (1988), and sporadic cases have been reported by others (Hanson and Rowland 1971, Taylor et al. 1982, Matsuzaka et al. 1986). It is possible that there is an autosomal recessive form of IFSH MD. Alternatively, lack of evidence of muscle disease in a parent may be explained by nonpenetrance, or, for sporadic cases, by a new mutation. Fitzsimons et al. (1987) have pointed out that in some individuals with autosomal dominant FSH MD, Coats' disease may be more apparent than muscle weakness. An ophthalmologic examination is therefore a useful adjunct to clinical assessment of family members for genetic counseling.

It is important to recognize the severity and the fairly rapid rate of progression of the infantile variant. Once diagnosed, the question arises as to what orthopedic interventions are warranted to help maintain shoulder function, ambulation, and spinal stability. Definitive answers cannot be given owing to the infrequent occurrence of the condition and its

recent recognition as a distinct entity, but treatment approaches can be discussed.

The one shoulder stabilization procedure done in our series was ineffective. In other patients the rapidly progressive overall disability led us to defer shoulder interventions. In view of the tendency of foot dorsiflexion weakness not to evolve into severe fixed equinovarus deformity, below-the-knee orthoses can be used to control the foot and ankle. An in-continuity heel cord lengthening, as described by Vulpus and Stoffel (Tachdjian, 1990), is warranted if the tightness of the achilles tendon is worsening.

The key factor limiting the ability to walk is the severe and progressive hip extensor muscle weakness, which is not currently amenable to effective bracing. Fixed flexion contractures of the hip do not warrant surgical correction while the patients are still ambulatory, because the risk of weakening functional capability is high.

Spinal bracing for the lumbar lordosis was attempted in two ambulatory patients, but it was ineffective and quickly abandoned. The lordosis is initially compensatory to help maintain stability in the face of marked gluteus maximus muscle weakness. Once the trunk is straightened in the brace, ambulation is either more difficult or not possible. Spinal orthoses will be most effective when the patient becomes wheelchair-bound. We currently use well-padded spinal orthoses plus wheelchair supports and belts when the patient becomes wheelchair bound to help counteract the severe lordosis. Spinal fusion may also be considered. We have not performed fusion because the patients have been either comfortable with orthoses and wheelchair-support systems or too severely impaired by pulmonary involvement at the time of surgical consideration.

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