CONGENITAL MUSCULAR DYSTROPHY

THE IMPORTANCE OF EARLY DIAGNOSIS AND ORTHOPAEDIC MANAGEMENT IN THE LONG-TERM PROGNOSIS

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Twenty-seven children with congenital muscular dystrophy have been reviewed. Eight presented with orthopaedic problems at birth and nineteen with varying degrees of muscle weakness. Eleven children who were extremely weak and were chairbound or needed assistance to walk developed marked contractures of the lower limb. Diagnosis in these children was late (mean age 6.6 years), and only one had had regular physiotherapy; four had been immobilised for treatment of dislocated hips. Successful surgical release and mobilisation has been achieved in only four. In contrast, seven extremely weak children diagnosed early (mean age 1.7 years) and given regular physiotherapy have minimal or no contractures. Five of these are now walking, two with calipers. Of eight children with milder weakness who walked independently by the age of two years, only one has developed contractures requiring operation. Overall, only two patients have progressive weakness, in sixteen power has improved and in eight the disorder has remained static. One infant is too young for the outcome to be assessed.

Of seven children with congenital dislocation of the hips, only one responded to abduction splints. In the face of severe weakness, operative intervention is probably best delayed until the child is old enough to be mobilised in calipers. In view of the relatively good prognosis for maintenance or improvement of muscle power in congenital muscular dystrophy, early diagnosis and regular passive movements to prevent severe contractures are important, particularly if walking is delayed by dislocation of the hip or by severe muscular weakness.

Congenital muscular dystrophy is a rare disorder characterised by muscular weakness and hypotonia from birth, often with associated deformities of the limbs and congenital dislocation of the hips. Muscle biopsy shows widespread dystrophic change with variation in the size of fibres, and excess fat and connective tissue. However, histochemistry and electron microscopy show no features of a specific myopathy (Dubowitz and Brooke 1973). The condition remains relatively static or may even improve with time despite the severity of the biopsy changes, and intelligence is usually normal (Dubowitz 1978).

Although reports of single cases resembling congenital muscular dystrophy can be traced back to the turn of the century (Howard 1908), it remains a rare condition with few reports in the British and American literature. It is possible that the condition goes undiagnosed due to inadequate investigation of children with arthrogryposis or with joint contractures. Large series have been reported from Japan (Fukuyama, Kawazura and Haruna 1960) where associated mental retardation is common. Smaller series of children with

normal intelligence have been reported from Finland (Donner, Rapola and Somer 1975) and Switzerland (Vassella, Mumenthaler and Rossi 1969) but give little guidance on management. In this review of twenty-seven patients we have studied the effect of delay in diagnosis on the development of contractures and the role of active intervention in improving the prognosis, especially the ability to walk.

METHOD

All children had been referred to the Muscle Clinic at Hammersmith Hospital between 1972 and 1977 for diagnosis or advice on management and were aged between two months and eighteen years at their last review. Twenty-six children had muscle biopsies compatible with a diagnosis of congenital muscular dystrophy (Fig. 1); one child with an affected brother did not have a biopsy. Examination included assessment of functional abilities, grading of muscle power on the Medical Research Council scale, measurement of contractures and radiographs of affected joints.

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Fig. 1

Biopsy from right deltoid muscle of the child in Figure 4, showing variation in the size of the fibres, internal nuclei and excess fat and connective tissue. (Haematoxylin and eosin, $\times 320$.)

RESULTS

Mode of presentation

Thirteen patients presented at birth, five being obviously weak and floppy and eight with orthopaedic problems (Table I). Of these eight, five were overtly weak at birth but in three cases weakness was not noticed until a later date. Twelve children presenting between six weeks and two years with delay in motor milestones were found to be weak, as were two children who presented with scoliosis at nine months and fourteen years respectively. Thus seventeen children were not noticed to be weak at birth by either their parents or attending physicians, but a careful history revealed that fifteen had had feeding difficulties or poor control of the head as neonates. In two children the first hint of any abnormality was failure

to crawl. Even after weakness was noticed there was often a further delay before referral for muscle biopsy, the mean interval between presentation and diagnosis being 5.1 years (range 1 month to 16.8 years). Weakness was generalised, being most severe proximally, and mild facial weakness was also common. The following extreme example illustrates how profound weakness could go unnoticed for several years.

A girl presenting with bilateral dislocation of the hips at birth was immediately put in abduction splints. Subsequently, the hips redislocated and she required a further operation. When at the age of five both hips were successfully contained she was found to be too weak to walk, but this was ascribed to her prolonged immobilisation. She was eventually referred for muscle biopsy at the age of eight years when she showed generalised weakness, severe contractures of the hips, the knees and the elbows, and a scoliosis of 24 degrees. Muscle biopsy showed the characteristic changes of congenital muscular dystrophy. Questioning the parents revealed that from birth she had always been generally inactive with poor control of the head.

Table I. Orthopaedic problems present at birth

"Arthrogryposis" with contractures of hips, knees, ankles, elbows, wrists and fingers	1
Bilateral dislocation of the hips and kyphoscoliosis	1
Unilateral dislocation of the hip and severe talipes equinovarus	1
Bilateral dislocation of the hips alone	1
Unilateral dislocation of the hip alone	3
Bilateral talipes equinovarus	1
No orthopaedic problems at birth	19
Total	27

Subsequent progress

One child (with arthrogryposis) is only two months old, so progress cannot be judged. Of the remaining twenty-six, only two have progressive weakness; muscle power has remained static in eight and actually improved in sixteen. There have been three deaths: one extremely weak chairbound child who died of pneumonia, and two ambulant adolescents, one with a rapidly progressive scoliosis who had a sudden respiratory arrest and the other dying in respiratory failure during an acute chest infection. On follow-up, the children fall readily into two groups: eighteen with severe weakness confined to wheelchairs or needing assistance to enable them to walk, and eight with milder weakness who all walked independently before two years of age.

Severe group. Eleven severely affected children developed contractures of the lower limb sufficient to interfere with walking (Fig. 2). Nine were confined to wheelchairs and two walked with a lot of help. Late diagnosis was common (mean age 6.6 years, range 3.5 to 9.8 years) and only one child had received regular physiotherapy; four had been immobilised during treatment for dislocated hips. In all cases contractures

Fig. 2

Fig. 3

Figure 2—A boy aged nine years who had dislocated hips and profound weakness at birth and was confined to a wheelchair. Figure 3—After bilateral tenotomies of the hips and knees and fitting of calipers the boy is standing for the first time.

were too severe or too longstanding to be relieved by passive stretching. Four children have had successful operative releases, one now walking independently, two walking in calipers and one standing in calipers (Fig. 3). Two more await operation. Of five who were thought unsuitable for operation, four appear to be getting stronger with time and could possibly have walked with or even without calipers had such severe contractures not been allowed to develop.

children in Cases 1 to 4 have had regular physiotherapy since infancy and have minimal contractures or none; three are now walking, one alone, one in long-leg calipers and one with a rollator frame; the youngest is not yet walking. The children in Cases 5 and 6 both had congenital dislocation of the hips with limited abduction, but with regular physiotherapy have no other contractures. The child in Case 5 has had an open reduction and derotation osteotomy and is now walking with a frame.

Table II. Outcome in seven severely affected children given physiotherapy

Case number	Age of diagnosis (years) (months)		Contractures	Age of walking (years) (months)		Walking aids
1		8	5 degrees of flexion of the hip	6	8	Frame None
2	1	6	10 degrees of flexion of the hips 5 degrees of flexion of the knees	4	6	Calipers
3		11	5 degrees of flexion of the hips	3	7	Frame
4	1	6	None	Not yet (2 10)		
5	2	10	Dislocated right hip with limited movement	2	10	Frame
6	1	1	Bilateral dislocation of the hips with limited movement	Not yet (1 10)		
7	3	11	45 degrees of flexion of the hips 30 degrees of flexion of the knees Responded to intensive physiotherapy	3	6	Calipers

Severe case referred late. An eight-year-old boy was referred to us for advice on management of severe contractures. He was born by extended breech delivery and was noticed to be hypotonic and weak at birth. At the age of six weeks a dislocated right hip was diagnosed and abduction splints applied. The hip redislocated when splints were removed at six months and the left hip was also found to be dislocated. Motor development was markedly delayed: he was unable to sit unaided or to support his head until one year. At two and a half years of age he had bilateral tendon transplants to stabilise his hips. Power in his arms and legs improved slightly and for several years he was able to stand with assistance, though he never walked and spent most of his day confined to a wheelchair. He gradually developed contractures of his knees, hips, elbows and shoulders. He had three episodes of pneumonia.

When seen by us at the age of eight years, he was an intelligent child with moderate weakness and severe contractures (Fig. 4). He had good power against gravity in all muscle groups but made poor effort against any resistance. He had flexion contractures of 30 degrees at both hips with a range of flexion and extension reduced to only 60 degrees. He also had 30 degrees of fixed flexion at both knees. It was considered that he probably had sufficient strength to have walked had contractures not developed, but an extensive operation would now be required to mobilise him. This was not undertaken and the child died at home of pneumonia six months later.

Seven extremely weak children who were diagnosed earlier (mean age 1.7 years, range 0.7 to 3.9 years) have had regular physiotherapy. Their clinical state is outlined in Table II. For each child, daily passive stretching of the hips, the knees and the ankles has been carried out by the parents, and two of the seven children have ankle splints to wear at night. In addition, these children have been treated actively by local hospital or school physiotherapists to encourage walking. The

Fig. 4

A boy aged eight years with severe weakness from birth, showing secondary contractures.

The girl in Case 6 is awaiting admission for reduction of her hips. The boy in Case 7 was first seen by us at the age of three and a half years when he had marked flexion contractures of the hip and the knee. He was admitted to

the ward for possible operation but intensive physiotherapy alone produced sufficient improvement to allow mobilisation in lightweight calipers. Eight of these eighteen children have developed a fixed scoliosis, most severe in the chairbound cases (Fig. 5).

Fig. 5

Spinal radiograph from an extremely weak boy with thoracolumbar scoliosis.

Severe case referred early. A boy aged eighteen months was referred because of inability to walk. He was the second of non-identical twins and at birth was noticed to be floppy and weak with a dislocated right hip. He was placed in abduction splints but each time these were removed the hip redischlocated. Open reductions at the age of eleven and fourteen months were only temporarily successful. He sat at thirteen months but remained unable to bear weight. When seen at the age of eighteen months he had a subluxed right hip (Fig. 6). He had 15 degrees of fixed flexion at both hips and elbows. His parents were taught to do passive stretching of these joints which they did daily and his contractures gradually resolved. At the age of two years seven months he had a rotation osteotomy of the right femur (Fig. 7) and was then given intensive physiotherapy. He has learned to walk with a rollator frame.

Milder cases. Eight children who were all walking by the age of two years were diagnosed as having congenital muscular dystrophy at the mean age of 9.7 years (range 1.2 to 18.8 years). One child developed bilateral equinus deformities before diagnosis and has had successful operative releases. Three other children had some tightness of their calcaneal tendons for which they are now having regular passive stretching. One developed a severe progressive adolescent scoliosis and died suddenly.

Dislocation of the hips

Seven children had dislocated hips at birth, four bilaterally and three unilaterally. All but one were born by the breech. In one child the hips were easily reducible and responded to conservative treatment in abduction splints. In three patients recurrent dislocation necessitated open reduction and derotation osteotomy and in three unoperated patients the hips remain dislocated. In one of our late-referred cases, the main hindrance to walking has been an untreated complete dislocation with marked associated contractures (Fig. 8).

DISCUSSION

In Britain congenital muscular dystrophy is rare or possibly underdiagnosed and in this series late diagnosis

Fig. 6

Fig. 7

Figure 6—Radiograph of a boy aged two years with repeated dislocation of right hip. Figure 7—Repeat radiograph after derotation osteotomy.

Fig. 8

Radiograph of the hip of the child shown in Figures 2 and 3.

was common. We have confirmed the finding of Donner *et al.* (1975) in Finland that joint contractures are a common complication, particularly if walking is delayed by severe weakness or by dislocation of the hip. In view of the good prognosis for power it is particularly important to preserve a full range of movement until the child can be mobilised. Ideally the diagnosis should be made in infancy and the parents taught how to do daily passive stretching exercises with regular supervision by a physiotherapist, night splints being fitted if tightness occurs. Active exercise to encourage mobilisation should also be given, but if the child remains too weak to bear weight then the use of calipers must be considered. Should joint contractures develop despite physiotherapy, as in one of our cases, then an active surgical approach is justified, as it may be possible to mobilise even severely weak children with the help of long-leg calipers. If an operation is undertaken the child should be got out of bed and mobilised in plaster casts the next day to prevent further loss of strength. In

neglected cases with severe contractures it may be difficult to assess the child's strength because of the limited range of movement. Moreover, tenotomies in themselves produce a slight loss in power, making prevention of contractures all the more important.

Scoliosis is also a common complication which may require spinal support and would probably be less severe if the children were mobilised early, spending the least possible time in a wheelchair. Even mild ambulant cases have a tendency to develop contractures and therefore require regular assessment. Furthermore, the condition is inherited as an autosomal recessive, so early diagnosis is essential if the appropriate genetic advice is to be given.

Children with dislocated hips at birth form a particularly difficult group to treat. Although a Von Rosen splint is worth trying initially, the hips may redislocate when the splint is removed. Prolonged immobilisation is probably not advisable in infancy in the face of underlying muscular weakness and it may be preferable to give daily physiotherapy to prevent contractures and delay operative intervention until the child is old enough and has enough strength to be mobilised in calipers after operation. In view of this difference in management from the conventional treatment in an otherwise healthy child, it is particularly important that strength and activity are assessed before a newborn infant with dislocated hips is put in splints. The possibility of an underlying myopathy should be seriously considered for any child whose hips repeatedly redislocate or any infant with multiple joint contractures. "Arthrogryposis" should be considered a symptom, not a diagnosis.

In conclusion, early diagnosis of congenital muscular dystrophy may considerably alter the management. A greater awareness is needed of the relatively good prognosis for power and the need to prevent the development of joint contractures, if the child's full potential for walking is to be achieved.

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REFERENCES

- Donner, M., Rapola, J., and Somer, H. (1975) Congenital muscular dystrophy: a clinico-pathological and follow-up study of 15 patients. *Neuropædiatrie*, **6**, 239-258.
- Dubowitz, V. (1978) *Muscle Disorders in Childhood*. London and Philadelphia: W. B. Saunders.
- Dubowitz, V., and Brooke, M. H. (1973) *Muscle Biopsy: A Modern Approach*. London and Philadelphia: W. B. Saunders.
- Fukuyama, Y., Kawazura, M., and Haruna, H. (1960) A peculiar form of congenital progressive muscular dystrophy. Report of 15 cases. *Pediatrics Universitatis Tokyo*, **4**, 5-8.
- Howard, R. (1908) A case of congenital defect of the muscular system (dystrophia muscularis congenita) and its association with congenital talipes equino-varus. *Proceedings of the Royal Society of Medicine*, **1**, Pathological Section, 157-166.
- Vassella, F., Mumenthaler, M., and Rossi, E. (1969) Congenital muscular dystrophy. In *Muscle Diseases*. Edited by J. N. Walton, N. Canal, and G. Scarlato. International Congress Series No. 199, pp. 620-623. Amsterdam: Excerpta Medica.