Adult onset scapuloperoneal myopathy

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SYNOPSIS Six cases are described of muscle weakness and wasting of scapuloperoneal distribution with an onset in early adult or middle life and a relatively benign progression. One case also showed mild facial weakness. Four cases were probably sporadic but in two, a mother and daughter, autosomal dominant inheritance was likely. Electromyographic studies demonstrated myopathic features in all, and this was confirmed by muscle biopsy in five. Electrocardiographic abnormalities were present in three cases, but their significance is uncertain. It is considered that adult onset scapuloperoneal myopathy constitutes a clinically distinct condition.

The unusual combination of muscular weakness affecting the upper limbs proximally and the lower limbs distally was noted at least as early as 1886 by Brossard. Since then several other reports have appeared, some describing affected families, although the nature of such cases remains the subject of discussion. The name 'scapuloperoneal amyotrophy' was tentatively proposed by Davidenkow (1927), who wrote extensively on the subject (see Davidenkow, 1939), but almost certainly a variety of disease entities is encompassed by this term. Apart from the distribution of the wasting and weakness, which typically affects the muscles of the neck, shoulder girdle and upper arm muscles, and the anterior tibial and peroneal muscles in the legs, other features may be encountered. The tendon reflexes are commonly absent and pes cavus, sensory disturbances, and cardiac involvement may occur. Considerable speculation has arisen as to the underlying mechanisms in cases displaying this unusual distribution of muscle weakness—in particular, whether a neurogenic or a primary myopathic process is implicated. The present report describes six cases with an onset of symptoms in adult life in which the evidence strongly favours a myopathic basis. They were either sporadic or of probable autosomal dominant inheritance.

FIG. 1 Case 1. The abnormal configuration of the shoulder girdle is well shown in (a) and the winging of the scapulae in (b).

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(Accepted 7 May 1975.)

CASE REPORTS

CASE 1 (Royal Free Hospital No. 1E28169) G.B., a 66 year old female with no family history of neurological disorder or parental consanguinity, first presented at the age of 48 years with difficulty in walking from bilateral foot drop. She was seen 15 years later, her gait having markedly deteriorated. Examination then demonstrated bilateral pes cavus and a harsh systolic murmur heard widely over the precordium without other features of cardiac disease; she was of low intelligence, although this was not assessed by formal psychometric testing. On neurological examination she showed slight bilateral facial weakness associated with a myopathic facies. There was striking weakness of neck flexion and weakness and wasting of the trapezii, rhomboids, spinati, pectoral and serratus anterior muscles, particularly on the right, with pronounced winging of the right scapula (Fig. 1). Minor weakness was apparent in the triceps and wrist extensors and the small muscles of the hand, again affecting the right arm more than the left. The trunk was normal. In the legs, there was slight bilateral weakness of hip flexion but she retained only a trace of ankle dorsiflexion and eversion. Plantar flexion and inversion remained only moderately impaired. Bilateral wasting of the anterior tibial and peroneal muscles was apparent. Sensory examination was normal. All tendon reflexes were diminished and the ankle jerks absent; the plantar responses were unobtainable.

The serum creatine kinase activity was slightly elevated (104 IU/l). Electromyography of the left tibialis anterior and right supraspinatus, rhomboid and deltoid muscles with a coaxial needle electrode demonstrated no spontaneous activity and a full

motor unit recruitment pattern in each of these muscles, with individual units which were brief and polyphasic. Some units of slightly increased amplitude (4–5 mV) were encountered in the rhomboids. No electrical activity could be demonstrated in the

FIG. 3 Case 2. The distal wasting in the lower legs with preservation of muscle bulk in the thighs is well shown in (a). Wasting of the shoulder girdle muscles and winging of the scapulae are evident in (b).

FIG. 2 Case 1. Transverse section from left triceps muscle. This shows an increased variability in muscle fibre size, with scattered atrophic fibres. There is some increase in endomysial connective tissue and the sarcolemmal nuclei are probably slightly more numerous than normal. Trichrome stain, $\times 120$.

right supraspinatus. Biopsy of the left triceps muscle (Fig. 2) revealed many randomly scattered, small, atrophic fibres, some of which were undergoing necrosis and phagocytosis, and there was a marked increase in endomysial connective tissue. On histochemical staining, both type I and type II muscle fibres were seen to be affected. Cardiac assessment revealed normal cardiac size radiographically, the electrocardiogram showed left anterior hemiblock, and an echocardiogram some thickening of the myocardial septum. The cardiac murmur was thought to represent mitral regurgitation and the asymptomatic cardiac abnormality, in association with a conduction disturbance, consistent with either ischaemic heart disease or a cardiomyopathy.

CASE 2 (Royal Free Hospital No. W5026) L.K., a 48 year old male previously in good health, developed difficulty in walking associated with mild bilateral foot drop at about the age of 40 years. A maternal uncle had some trouble with both legs but no further details were obtainable. The weakness slowly increased until severe bilateral foot drop had developed, associated with marked distal wasting (Fig. 3) and complete selective paralysis of the anterolateral muscle groups in the legs with sparing of the small muscles of the feet. Examination also revealed asymptomatic bilateral wasting and weakness of the scapular and upper arm muscles with sparing of the forearm, sternomastoid and trapezius muscles. The subsequent progression of the disease resulted in difficulty in elevation of the arms above the head and a marked 'steppage' gait. Repeated examinations over eight years have revealed further weakness of his scapular and upper arm muscles with slight weakness and wasting in the small muscles of the hands and some wasting of both trapezii. The involvement of the legs and slight weakness of ankle plantar flexion has similarly increased slightly. At no time have there been sensory or sphincter disturbances. The reflexes have been absent apart from bilateral diminished knee jerks; the plantar responses have remained flexor. General examination has revealed no abnormality.

Investigations in 1966 when the patient was aged 41 years revealed a slightly elevated serum creatine kinase activity (92 IU/l), and electromyography of the right infraspinatus, supraspinatus, biceps, and peroneus longus muscles demonstrated features typical of a myopathy. Biopsy of the left tibialis anterior muscle demonstrated severe changes. There was a considerable increase in collagenous and fatty endomysial tissue. Muscle fibre diameter was abnormally variable, with some fibres of increased size and others of reduced diameter. There were excessive numbers of centrally situated sarcolemmal nuclei.

L.K. was further investigated in 1969 when the serum creatine kinase activity was within normal limits. An ECG demonstrated left axis deviation with deep Q waves in leads II, III, and AVF with a QRS duration within the normal range at 0.08–0.10 seconds. Recently performed quantitative electromyography (Dr R. G. Willison) with concentric needle sampling of the right triceps muscle showed abnormalities consistent with a myopathy: against a fixed 2 kg weight, a mean number of 603 turns/second of mean amplitude of 0.33 mV were recorded (see Rose and Willison, 1967).

CASE 3 (Royal Free Hospital No. U10725) R.T., male aged 67 years without significant family history, first noticed muscle weakness in the legs when aged about 28 years and was told that he must have had

FIG. 4 Case 3, showing pronounced distal wasting in both lower legs.

poliomyelitis. Ten years later, however, he presented with progressive difficulty in walking because of bilateral foot drop. More recently, he had noticed some mild weakness of the upper arms. At the age of 58 years (Fig. 4), his abnormal signs consisted of minimal wasting of the scapular muscles with moderate weakness of the spinati, deltoid, and triceps muscles bilaterally. There was gross wasting and weakness of all the distal muscles in the legs especially of the tibialis anterior; little movement was possible at the ankles and this was confined to inversion and plantar flexion. The extensor digitorum brevis muscles, however, were bulky (Fig. 5). Sensory examination was normal as were the tendon reflexes in the upper limbs, but the knee jerks were reduced and those at the ankle absent; the plantar responses were flexor. Mild bilateral pes cavus was present and he had a lordotic stance and an unsteady 'steppage' gait. He has been followed up for over 10 years and has had a very slowly progressive disability with increasing weakness now affecting his trunk and he walks with the aid of two sticks.

Investigations when aged 58 years revealed markedly elevated serum creatine kinase activity (1 280 IU/l). Electromyography with sampling of the right rectus femoris and left tibialis anterior muscles showed a slightly reduced motor unit recruitment pattern, individual motor unit potentials being brief, poly-

phasic, and of low amplitude. Sampling of the left extensor digitorum brevis demonstrated no abnormal insertion activity or spontaneous electrical activity, but the number of motor units under voluntary control was reduced. In this muscle, some individual motor unit potentials were of increased size (5–10 mV). Sampling of the left vastus lateralis revealed normal findings and motor nerve conduction velocity in the left peroneal nerve was also normal (42 m/s). Quantitative electromyography (Dr R. G. Willison) was normal in the left quadriceps and triceps muscles, but sampling of the right quadriceps showed a definite abnormality with counts up to 900/s with a mean amplitude of 0.1 mV at 5 kg tension. Biopsy of the left tibialis anterior muscle demonstrated gross abnormalities. Muscle fibre diameter showed considerable variability with fibres both of increased and reduced size. There was an increased number of centrally situated sarcolemmal nuclei, and evidence of fibre splitting. Fatty and collagenous connective tissue was increased in amount. The electrocardiogram was normal. The electrodiagnostic and histological investigations thus demonstrated a myopathic process without evidence of peripheral nerve dysfunction, except for some features suggestive of chronic partial denervation in the left extensor digitorum brevis muscle.

CASE 4 (Royal Free Hospital No. 1E31028) L.G., a 67 year old male, presented with a 10 year history of bilateral weakness of both feet resulting in foot drop. He had undergone an arthrodesis of both ankles in 1971 with only temporary improvement in his gait and recently he had had to use crutches for walking. Over the previous five years he had developed progressive weakness of both arms, mainly affecting the shoulders and upper arms, but had retained good function in his hands. He had a three year history of hypertension, recurrent left ventricular failure, and chronic obstructive airways disease. His family history indicated that one of two brothers died aged 62 years with heart disease but without neurological disability; his father died at the age of 91 years and had had a long-standing disturbance of gait which was never investigated. Recent neurological assessment of the patient revealed moderate wasting of both trapezii. The scapulae were laterally placed and showed 'winging' on elevation of the arms. The rhomboids and serratus anterior were weak bilaterally, as were the deltoids, pectoral, and triceps muscles. Apart from very mild weakness of finger extension and abduction, the forearm and hand muscles were normal. In the legs, pes planus and enlargement of both ankle joints were noted. There was bilateral wasting of thigh and anterior tibial muscles, but no weakness of the hip and thigh

FIG. 5 Case 3, showing preservation of the extensor digitorum brevis muscle (arrowed).

muscles was detected. Dorsiflexion at the ankles and extension of the toes were absent bilaterally, and ankle eversion was grossly weak. Inversion and plantar flexion at the ankles, despite the previous arthrodesis, were only slightly impaired, as was flexion of the toes. All tendon reflexes were sluggish and the plantar responses were flexor; sensory examination was negative. At the time of examination, the patient having recently been discharged from another hospital, there was no evidence of heart failure, although his pulse remained irregular.

Investigations revealed serum transaminase values to be within the normal range, but the serum creatine kinase activity was slightly elevated (142 IU/l) when no longer in cardiac failure. Electromyography performed on the left triceps and supraspinatus muscles showed no spontaneous activity, a diminished recruitment pattern with motor unit potentials of brief duration and small amplitude. No electrical activity was elicited in the left tibialis anterior or peroneal muscles. Median nerve motor conduction velocity was normal (67 m/s), as was an index finger/wrist median sensory action potential. The electrocardiogram demonstrated atrial fibrillation and evidence of anterolateral myocardial ischaemia. A muscle biopsy taken at another hospital from the left forearm flexor group showed some increase in variability in muscle fibre size, and abnormal numbers of centrally situated sarcolemmal nuclei.

CASE 5 (Royal Free Hospital No. 1E27894) M.P.,

a female now aged 27 years, was well until the age of 18 years when she began to catch the toes of her left foot, and a similar disability soon affected her right foot. This disability gradually increased, she developed marked bilateral foot drop and within eight years had become unable to run. She noticed no weakness of her upper limbs. Her mother (case 6) is affected by a similar disorder. There is no parental consanguinity and no other history of neuromuscular disease in the family. Examination of the cranial nerves and upper limbs was normal except for bilateral wasting of the rhomboids and mild weakness of the deltoid muscles. In the legs, bilateral wasting of the anterolateral group of muscles was present and there was marked bilateral weakness of ankle dorsiflexion and eversion and of toe extension. The extensor digitorum brevis muscles were normal. Fasciculation was not seen and sensory examination was normal. All the tendon reflexes were present, except for those at the ankle; the plantar responses were flexor. She walked with a 'steppage' gait.

Investigations showed an elevated serum creatine kinase activity (200 IU/l). Electromyography (Dr R. G. Willison) performed on the right tibialis anterior muscle demonstrated no spontaneous activity; on voluntary contraction no detectable force was developed, but a few motor units were seen discharging at high rates and their action potentials were brief and polyphasic with brief spikes up to 3 mV in amplitude. The electrocardiogram was normal. Biopsy of the extensor hallucis and tibialis anterior muscles (Fig. 6)

FIG. 6 Case 5. Transverse section from right tibialis anterior muscle. This shows fatty replacement and considerable variation in muscle fibre size. Central nuclei are common. Trichrome stain, $\times 190$.

showed considerable loss of muscle fibres with fatty connective tissue replacement. There was marked variation in muscle fibre size, numerous small and often grouped, round fibres together with normal and hypertrophied ones. Centrally placed sarcolemmal nuclei, hyaline degeneration, occasional basophilic change and necrosis with macrophage response were also seen. The changes affected both type I and II fibres, which showed a normal mosaic pattern.

CASE 6 D.W., a female, the mother of case 5 and now aged 54 years, developed very mild bilateral foot drop during adolescence. Although the disability very slowly progressed, it resulted in insignificant disability until she was aged about 40 years when she began to trip easily. Since then, she has become aware of gradually increasing weakness of ankle dorsiflexion but is able to lead a normal life and has not required medical attention. Examination now reveals prominently sloping shoulders and bilateral slight wasting and weakness of the deltoid muscles. There is thinning of the muscles of the anterolateral compartment of the lower limbs with very gross bilateral weakness of ankle dorsiflexion and minimal eversion, but normal inversion and plantar flexion. Minimal extension of the toes is possible but flexion is normal. The trunk and face are spared and sensation is normal. The tendon reflexes also are unremarkable except for absent ankle jerks and the plantar responses are unobtainable. She walks with a bilateral foot drop and 'steppage' gait. Limited investigations only were possible: electromyographic findings (Dr R. G. Willison) were found to be similar to those obtained in case 5, although of slightly greater severity; her electrocardiogram and the serum creatine kinase level were normal.

DISCUSSION

Despite the fact that patients exhibiting muscle weakness and wasting with a proximal distribution in the upper limbs and a distal distribution in the lower were first described about a century ago (Brossard, 1886), the nosology of the scapuloperoneal syndrome remains confused. It seems clear that the syndrome encompasses a number of different genetic entities. Part of the difficulty stems from the fact that laboratory studies have not always been successful in establishing whether the underlying process is myopathic or neurogenic. This has been our own experience and was emphasized, in particular, by the series of cases reported by Feigenbaum

and Munsat (1970). Mixed or inconclusive changes have also been documented by Kaeser (1965), Ricker *et al.* (1968), and Takahashi *et al.* (1974). When the diagnosis has to be made in the absence of necropsy studies, the interpretation of muscle biopsy appearances can be complicated by the presence of secondary myopathic features (Drachman *et al.*, 1967), and it is possible that 'pseudomyopathic' appearances could appear in the electromyogram if individual branches of the terminal arborization of motor axons were selectively affected (Kaeser, 1965). Furthermore, serum creatine kinase levels may be elevated both in myopathic disorders and in conditions that give rise to chronic denervation atrophy (Williams and Bruford, 1970).

Notwithstanding these qualifications, it seems evident that some examples of the scapuloperoneal syndrome have shown undoubted features of denervation electromyographically (Emery *et al.*, 1968; Zellweger and McCormick, 1968; Meadows and Marsden, 1969; Caraceni *et al.*, 1972) and in muscle biopsies (Emery *et al.*, 1968; Meadows and Marsden, 1969). Motor nerve conduction velocity has been recorded as being normal or occasionally somewhat reduced (Emery *et al.*, 1968). In one patient, described by Meadows and Marsden (1969), a progressive diminution in motor conduction velocity over a 10 year period was documented. The absence of sensory loss and the relative preservation of motor nerve conduction velocity has led to these cases being classified as examples of 'spinal muscular atrophy'. It was considered that the degeneration of anterior horn cells and of cranial nerve motor nuclei found in the necropsy study reported by Kaeser (1965) supported this contention, although a muscle biopsy from an affected member of the same family was suggestive of myopathy.

Some cases of the scapuloperoneal syndrome have been found to have distal sensory loss in the limbs (Davidenkow, 1939), and to show abnormalities of sensory nerve conduction (Thomas, 1975, unpublished observation), indicating that at times the syndrome may involve both the lower motor and first sensory neurones. Adequate studies of sensory nerve conduction have not been performed in all instances where a diagnosis of 'spinal muscular atrophy' has been made; they are mandatory before a state-

ment that the disorder is confined to the lower motor neurones can be proposed.

In a third category of case, myopathic changes have been found, electromyographically (Seitz, 1957; Hausmanowa-Petrusewicz and Zielińska, 1962; Ricker and Mertens, 1968; Lovelace and Menken, 1969; Serratrice *et al.*, 1969; Rothauwe *et al.*, 1972), and at necropsy (Thomas *et al.*, 1972).

In the myopathic form of the scapuloperoneal syndrome, sporadic, autosomal dominant, and X-linked cases have been described. Families displaying dominant inheritance will be discussed later. Rothauwe *et al.* (1972) reported a family in which 17 males in three generations were affected, the inheritance being of X-linked pattern. The disorder began in childhood with muscle contractures that gave rise to limitation of neck and elbow flexion, and also with contractures of the calves. This was associated with weakness of a scapulohumeroperoneal distribution. Cardiac arrhythmias were an important feature, and nine cases died suddenly between the ages of 37 and 59 years. The disorder in the family reported by Thomas *et al.* (1972) was closely similar and showed linkage with deutan colour blindness.

It is considered that the cases reported in this communication, on the basis of electromyographic studies and on muscle biopsy when this could be undertaken, show features highly suggestive of a myopathic disorder. They are all characterized by an onset of symptoms in young adult or middle life and a relatively benign progression. In none is the patient wheelchair bound or grossly incapacitated, although three are now in their seventh decade. In each, symptoms in the legs were the initial presentation and difficulty in walking because of bilateral foot drop was a prominent feature. Symptoms from weakness of the shoulder girdle and upper arm muscles became manifest after those in the legs. Apart from the scapuloperoneal distribution of the muscle weakness, the facial muscles were affected in one case. Muscle pseudohypertrophy was possibly present in one instance. A striking feature was the sparing of the small muscles of the feet. The extensor digitorum brevis was not involved, even in cases with gross weakness and wasting of the anterolateral compartment muscles of the lower legs. This is in marked contrast with the pattern

of involvement of the legs in peroneal muscular atrophy or spinal muscular atrophy of distal distribution, where early involvement of the small foot muscles is the rule (Panayiotopoulos and Scarpalezos, 1975). Pes cavus was present in two cases in the present series, pes planus in one. Muscle fasciculation and sensory abnormalities were never encountered; the tendon reflexes were reduced or absent. In three patients, the electrocardiogram was abnormal. Although the abnormalities would be consistent with a cardiomyopathy, at least in cases 1 and 2, at the ages at which the electrocardiographic abnormalities were noted, without further cardiological features or additional detailed investigation, it would not be possible to exclude other causes such as ischaemic heart disease.

Cases 1 and 3, in the absence of any clear family history of similar disorder, must be considered to be sporadic examples of the disease. In cases 5 and 6, autosomal dominant inheritance is likely. In cases 2 and 4, the nature of any hereditary factors must remain undetermined.

The present series of cases resembles most closely those reported by Ricker and Mertens (1968), Lovelace and Menken (1969) and Serratrice *et al.* (1969). In none of these reports are cardiac features discussed. Ricker and Mertens (1968) reviewed the histories of 212 patients with muscular dystrophy seen over a 15 year period. Thirteen cases were found that accorded with the scapuloperoneal syndrome. Of these 13 patients, five were members of three families in which successive generations were affected, and in 10, electromyographic and muscle biopsy confirmation was obtained. In 12 of the 13 cases the initial involvement was of the upper limb and scapular muscles, contrasting with the early distal involvement in the legs observed in the present series. Interestingly, it was noted that later involvement of the pelvic girdle, thigh, and trunk muscles occurred, reminiscent of our case 3. Lovelace and Menken (1969) described three male cases with an onset in adolescence or early adult life and slow progression. Two presented with foot drop, one with winging of the scapulae. Electromyography revealed myopathic changes and muscle biopsy in all three indicated primary muscle disease. The serum creatine kinase activity was persistently elevated in two. Serratrice *et al.* (1969) reported 14 cases of the scapuloperoneal

syndrome in nine of which autosomal dominant inheritance was suggested, although detailed genetic studies were not undertaken. Facial involvement was observed with some frequency, and it is of interest that, in two families, two siblings of the same sex and approximately the same age were identified, in only one of whom was there facial involvement.

Of the cases reported by Ricker and Mertens (1968), one also had generalized neurofibromatosis; another had Klinefelter's syndrome, confirmed by testicular biopsy. The authors discussed at length the relationship of the scapuloperoneal syndrome to other myopathic disorders, and the possibility of the evolution of the syndrome into other better recognized myopathies. The clinical features of the present series, however, clearly indicate that they represent a distinct group. It is as yet uncertain whether they are themselves genetically homogeneous.

We wish to thank Mr Lipmann Kessel for referring case 1, Dr Nigel Compston for case 4, and Professor John Marshall for cases 5 and 6. We are indebted to Miss J. M. Workman for technical assistance and to the Wellcome Trust and the Muscular Dystrophy Group of Great Britain for financial support.

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