Spinal muscular atrophy in childhood

Two clues to clinical diagnosis

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Moosa, A., and Dubowitz, V. (1973). *Archives of Disease in Childhood*, 48, 386. **Spinal muscular atrophy in childhood: two clues to clinical diagnosis.** A coarse tremor was noted in 13 children suffering from the childhood form of spinal muscular atrophy. Tremor has not been seen in any other condition producing proximal muscle weakness in childhood, and its presence should therefore suggest the diagnosis of spinal muscular atrophy.

In addition, the feet of patients with spinal muscular atrophy tended to evert, whereas in Duchenne muscular dystrophy there was either no deformity or a tendency to toe walking.

The child with a waddling gait who 'climbs up himself' by the Gowers manoeuvre, suggests a diagnosis of muscular dystrophy. Other conditions, with a similar distribution of weakness, can however produce a very similar clinical picture. 'All that waddles is not dystrophy' (Dubowitz, 1965).

The condition most commonly confused with muscular dystrophy is the benign variant of spinal muscular atrophy. The severe infantile form of spinal muscular atrophy (Werdnig-Hoffmann disease) has been recognized for a long time and is easily diagnosed. It is only recently, however, that more benign forms of this disease have been recognized, compatible with survival into adolescence and adult life (Byers and Banker, 1961; Dubowitz, 1964a). The most benign form is the Kugelberg-Welander syndrome (Wohlfart, Fex, and Eliasson, 1955; Kugelberg and Welander, 1956).

Diagnosis of these conditions is important because the prognosis as well as the pattern of inheritance is different. Duchenne dystrophy is usually inherited by an X-linked mechanism and its progression is relentlessly downhill. Spinal muscular atrophy, on the other hand, is inherited as an autosomal recessive character, and tends to run a relatively benign course. It usually remains relatively static, and some cases may even show some improvement in function.

One clinical feature which unequivocally separates spinal muscular atrophy from muscular dystrophy

is fasciculation of muscle; but this is not always present. It is more commonly seen in the tongue than in skeletal muscles (Dubowitz, 1964a; Munsat *et al.*, 1969).

In our experience two additional clinical signs—tremor and foot posture—often provide a clue to the diagnosis of spinal muscular atrophy, which can subsequently be confirmed by special investigation such as electromyography and muscle biopsy.

Results

Tremor. Over the past 3 years we have seen 13 patients with spinal muscular atrophy of the more benign childhood type (Table) all of whom had a coarse irregular tremor of the hands. In most of them the tremor was only observed when it was specifically looked for, but in two children it was quite obvious and was commented on by the parents—'the child is ever so nervous'.

In the older children it was easily demonstrable when the arms were stretched forward and the fingers splayed out. In the younger children it was readily seen when the patient built a tower of bricks. In most of the children the tremor was confined to the hands, but in some it also affected the head and other parts of the body. In only 4 children was there fasciculation of the tongue and/or of the limb muscles at the time the tremor was first noted.

It was not possible to assess the age of onset of the tremor as most parents were not aware of its presence. In some patients it was noted at the time of the first examination, while in others it was

TABLE
Tremor and foot deformity in children with spinal muscular atrophy

Case no.	Sex	Present age (yr)	Clinical status	Tremor		Age of onset of weakness (mth)	Fasciculation and atrophy of tongue	Fasciculation of other muscles	Posture of feet
				Noted at age of (yr)	Still present at (yr)				
1	M	6	Ambulant	4		13			Eversion
2	F	7½	Ambulant		7	12	+		Eversion
3	F	10½	Ambulant	8½	10½	Birth			Eversion
4	M	3½	Ambulant	1½	3½	9			Eversion
5	F	13½	Chair-bound	6	12	18			Eversion
6	M	8	Ambulant		8	6-12	+	Deltoid	Eversion
7	M	7½	Ambulant	4*	7½	24			Eversion
8	F	7	Ambulant	4		12			Limitation of dorsi-flexion
9	F	13	Chair-bound	1	13	12	+		Eversion
10	F	5	Ambulant	4	4½	11			Limitation of dorsi-flexion
11	M	6	Ambulant	4½	5	18			Eversion
12	F	5½	Ambulant	4	5	Birth			Eversion
13	F	5½	Ambulant	1½*	5	16		Deltoid	Eversion

*Noted by parents.

only noted on subsequent examinations. In one patient it was still present 12 years after it was first noted. The amplitude of the tremor varied from patient to patient and even in the same patient was influenced by such factors as fatigue and anxiety, which tended to accentuate the tremor.

We have not seen this tremor in any other patients with proximal muscle weakness due to conditions such as Duchenne dystrophy, congenital myopathy, and dermatomyositis.

Foot deformity. Another clinical sign we have found useful in distinguishing between patients with Duchenne muscular dystrophy and spinal muscular atrophy is the type of deformity of the foot that tends to develop in these patients.

In most of our patients with spinal muscular atrophy there was a tendency to walk flat-footed and for the feet to evert (Fig. 1). On the other hand, patients with Duchenne dystrophy either showed no deformity or tended to walk on their toes, with a tendency to equinovarus deformity of the feet (Fig. 2). This distinction, however, did not apply in children who had gone off their feet, after which most of them developed equinovarus deformities irrespective of underlying pathology, unless this was prevented by adequate postural support.

Discussion

Several reports in the literature have mentioned the presence of tremor in occasional patients with

Fig. 1

Fig. 2

FIG. 1.—A 5-year-old girl with spinal muscular atrophy. Note the everted posture of the feet.

FIG. 2.—A 10-year-old boy with Duchenne dystrophy. Note tendency to spontaneous equinus posture of feet.

spinal muscular atrophy. Brandt (1950) described one child who had universal trembling and mentioned that fasciculate tremor was seen more often when the disease began after the age of 1. Kugelberg and Welander (1956) mentioned one patient who had a fasciculate tremor of the toes. Byers and Banker (1961) noted the occurrence of tremor in the mildly affected patients. Dubowitz (1964b) described a patient who had 'voluntary fasciculation' of a finger. Spiro, Fogelson, and Goldberg (1967) and Peters *et al.* (1968) also mentioned the presence of tremor in their patients. One of the patients reported by Gardner-Medwin, Hudgson, and Walton (1967) had a 'familial tremor', and several members of the family with bulbospinal muscular atrophy described by Kennedy, Alter, and Sung (1968) had an 'essential' tremor of the hands.

Although there is frequent mention of this sign in the literature, its usefulness to the clinician faced with the problem of diagnosis in children with muscle weakness has until recently not been emphasized. Spiro (1970), under the formidable title of 'Minipolymyoclonus', drew attention to the usefulness of this sign in the diagnosis of spinal muscular atrophy. He did not think it was a true tremor as it was nonrhythmic, nor did he think it was due to fasciculation since twitching of a muscle fasciculus was not observed.

During studies in this department on the effect of denervation in mice (M. Strugalska, personal communication, 1971), a coarse tremor was noted in the denervated limb several weeks after denervation, at a time when reinnervation was occurring. This suggests that the tremor seen in patients with spinal muscular atrophy might possibly have a similar basis and be due to reinnervation. In support of this explanation is the absence of tremor in our cases of severe Werdnig-Hoffmann type of spinal muscular atrophy. Moreover, histochemical studies of biopsies from these patients show extensive fibre-type grouping in the more benign forms of disease and not as a rule in the severe infantile spinal muscular atrophy. The presence of fibre-type grouping is indicative of reinnervation of muscle.

Whatever the nature of this tremor, our observations parallel those of Spiro (1970) and indicate that tremor of the hands is a useful diagnostic sign in patients with proximal weakness, and clearly

separates those with spinal muscular atrophy from those with muscular dystrophy.

The tendency to equinovarus deformity of the feet in patients with Duchenne muscular dystrophy is thought to be due to imbalance of muscle action round the ankle joint, with the gastrocnemius exerting a more powerful action against the weaker anterior tibial muscles. On the other hand, the tendency to eversion in patients with spinal muscular atrophy is probably due to the hypotonia that usually accompanies the disease.

The presence of a tremor of the hands as well as a tendency to eversion of the feet should, therefore, alert one to the diagnosis of spinal muscular atrophy rather than muscular dystrophy.

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