Myelopathy in Marfan's syndrome

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SUMMARY A patient with Marfan's syndrome and a myelopathy is reported, and the association of multiple spinal arachnoid cysts noted. It is proposed that the basic connective tissue defect in Marfan's syndrome may predispose to the formation of arachnoid diverticuli and that in this case spinal cord damage was the sequel.

In 1896 Antoine Bernard-Jean Marfan described the skeletal features of the syndrome which now bears his name (Marfan, 1896). Weve (1931) coined the term *dystrophia mesodermalis congenita* and determined the autosomal dominant pattern of inheritance of this condition. Any contemporary account of the syndrome must acknowledge the tremendous store of information detailed by McKusick in his book on heritable connective tissue disorders (McKusick, 1972). These authors and many others have described the cardinal involvement of skeletal, ocular, and cardiovascular systems but published reports contain little reference to neurological disturbances. In the present report we describe a case of myelopathy complicating Marfan's syndrome, discuss the possible pathogenesis, and review briefly the subject of neurological involvement.

Case report

A 36 year old housewife (VN) has Marfan's syndrome as evidenced by ectopia lentis, iridodonesis, cataracts, rotary nystagmus, arachnodactyly, dolichocephaly, gothic palate, mitral systolic murmur, and the strong family history illustrated in Fig. 1. In 1976, she developed moderate difficulty in walking with weakness and a "leadens heaviness" of the lower limbs and foot drop on exercise. There was little progression of this disability until mid-1977 when she fell down stairs. After this she had a constant dull ache in the lower thoracic region and could only walk with the support of furniture. Her walking disability progressed so that by early 1978 she was unable to rise from her bed or chair without help. She had a mild numbness of her

Fig. 1 Family tree of the case illustrated—
■ = affected male, □ = unaffected male, ▣ = probably affected male, ● = affected female, ○ = unaffected female, ● = probably affected female. None of the children have neurological abnormality.

toes and urgency of micturition which had preceded her fall. Examination showed the features of Marfan's syndrome detailed above. There was a thoracic kyphoscoliosis and an angulated deformity of the upper lumbar spine. Neurological examination revealed brisk reflexes in the otherwise normal upper limbs. In the lower extremities there was a spastic paraparesis with brisk reflexes, sustained ankle clonus, and extensor plantar responses. An indistinct sensory level was detected at L 1 dermatome.

Routine investigations were unremarkable. The electrocardiograph showed a P mitrale configuration and the chest radiograph an unfolded aorta. Plain radiology of the spine confirmed the abnormality detected clinically with a wedged deformity involving L 2-3 vertebrae (Fig. 2). The spinal canal was widened throughout but the skull radiograph was normal with no evidence of basilar invagination. Cisternal myelography with 6 ml of iodophendylate demonstrated numerous large communicating sacral arachnoid cysts (Fig. 3). Similar cystic lesions were noted in the thoracic region by the neuroradiologist during screening but attempts

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Accepted 28 August 1978

Fig. 2 Lateral view of spine showing deformity at midlumbar region.

Fig. 3 Myelogram view showing sequestration of dye in communicating sacral cysts.

to delineate these further were unsuccessful because of sequestration of the dye in the sacral cysts. No further information was gained by rescreening 24 hours later.

A surgical approach was considered undesirable in view of the presence of multiple and widespread lesions, and conservative treatment with intensive physiotherapy was applied, with a moderate but worthwhile improvement in mobility.

Discussion

A case of Marfan's syndrome associated with a myelopathy and arachnoid cysts is presented. Trauma appears to have exacerbated the problem but spinal cord damage was present before this. The site of this damage cannot be placed precisely, and although the brisk arm reflexes suggest the possibility of some cord involvement at the cervical level, the major lesion is probably at low dorsal level. The absence of obstruction to the caudal flow of dye at myelography suggests that a gross mechanical block associated with spinal deformity or trauma was not present, and it is reasonable to postulate that the association of arachnoid cysts and myelopathy in this patient is not fortuitous.

Spinal cord compression is a well-recognised complication of arachnoid cysts (Raja and Hankinson, 1970; Palmer, 1974), and arachnoid cysts have been noticed previously in Marfan's syndrome (Mitchell *et al.*, 1967).

The definitive abnormality of connective tissue which results in the clinical features of Marfan's syndrome has yet to be determined. It is possible that the connective tissue investing the spinal cord in the form of the meningeal membranes is involved in the mesodermal dystrophy, and we suggest that this may be the case more frequently than clinical (McKusick, 1972) or negative pathological (Roark, 1959) studies would indicate. Indeed, enlargement of the spinal canal (Nelson, 1958) and scalloping of vertebrae (Mitchell *et al.*, 1967), have been reported previously in Marfan's syndrome, and the underlying cause may well be dural ectasia and associated arachnoid diverticuli.

Neurological manifestations of Marfan's syndrome are uncommon. Hypotonia is a frequent finding but this is caused by laxity of joints, tendons, and ligaments. It is this mechanical deficiency that is considered to explain the myopathy rarely observed in Marfan's syndrome (Goebel *et al.*, 1973), and there is no convincing evidence of

any specific primary muscular pathology. The hypotonia may, however, be so striking as to cause diagnostic confusion with the primary muscle disorders, particularly where spinal deformity is prominent (McKusick, 1972). Neural deafness occurs with significant frequency, 3% according to one author (Rados, 1942), but the reason for this is unclear. Isolated reports have appeared associating Marfan's syndrome with encephalocele, torticollis, carotid aneurysm (McKusick, 1972), internal hydrocephalus (Pasachoff *et al.*, 1944), diabetes insipidus (Halle and Collipp, 1971), basilar invagination (Yajnik *et al.*, 1975), and the interesting association of ectodermal abnormalities such as neurofibromata with mesodermal characteristics of the syndrome has been noted (El-Defrawi and El-Shewi, 1974). All these associations, including the present case report, await the ultimate unravelling of the biochemical and embryological defects of this condition.

We thank Dr Ronald Bruce for his help with the radiology of this case.

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