FRIEDREICH'S ATAXIA AND JUVENILE DIABETIC JOINT CONTRACTURE

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INTRODUCTION
There is a prevalence of insulin-dependent diabetes mellitus in Friedreich's ataxia ranging from 8 to 20 per cent.1 2 The occurrence of limited joint mobility in juvenile diabetes was first described in 19743 and subsequently has been identified in 50 percent of children after nine years of diabetes.4 We describe a case in whom joint contracture was discovered in a patient with a non-progressive form of Friedreich's ataxia. To our knowledge, this association has not previously been reported.

CASE REPORT
A 19-year-old girl with Type I diabetes mellitus of 15 years duration was admitted to hospital for investigation of severe ataxia, the onset of which was first noted at age 16. Growth rate had been slow, but development normal, with menarche at age 15. Both parents were healthy. However, of four siblings, a brother had Type I diabetes mellitus of 20 years duration complicated by retinopathy, neuropathy and nephropathy, and subsequently died of uraemia at the age of 32. He had no known evidence of limited joint mobility. Of three sisters, one had congenital adrenal hyperplasia, one had died at age four following surgery on transposition of the great vessels, and one was alive and well with normal results of neurological examination.

The patient was of short stature, 137.5 cm, weighed 41 kg, but was normally developed. She had bilateral pes cavus with minimal equinovarus deformity. There was grade II/VI systolic ejection murmur along the left sternal border but no electrocardiographic abnormalities. Fundoscopic examination revealed mild bilateral background retinopathy with early neovascularization in the area of the right disc and macula. She had episodic proteinuria, but blood urea and creatinine were normal. There was interphalangeal joint flexion deformity of both hands resulting in limited mobility, but without muscle atrophy, altered sensation or palmar fascial thickening. The skin of the hands appeared thickened. There was also limitation of movement at the knee and ankle joints. These findings are characteristic of the limited joint mobility seen in diabetes.

Neurological examination revealed ataxic gait, positive Romberg's sign and bilateral horizontal nystagmus. Muscle strength, sensation and reflexes were normal in the upper extremities, but there was diminished power of dorsiflexion at both ankles, absent tendon reflexes and posterior column and light touch sensory loss in the lower extremities. Babinski's sign was negative.

She is now 25 years old and has shown no deterioration of her neurologic status over the past six years. Her joint abnormalities have also remained stable and there has been no further progression of her retinopathy or nephropathy.
DISCUSSION

Friedreich's ataxia, the most common of the spinocerebellar degenerations, is an autosomal recessive disorder with childhood onset of progressive ataxia, absent deep tendon reflexes, long motor tract signs and posterior column sensory deficits. Pes cavus, kyphoscoliosis, nystagmus, dysarthria and cardiomyopathy are common. This patient was diagnosed at the age of 19 years as an incomplete form of the syndrome. Friedreich's ataxia has been associated with both Type I and Type II diabetes mellitus. More than two-thirds of the population with combined disorders have been young females with Type I diabetes as is this patient. Usually the ataxia is well advanced before the onset of diabetes. The present case is unusual in that the diagnosis of diabetes preceded the ataxia by 12 years. We are aware of only one other case in which diabetes preceded the neurological disorder.

Joint contracture due to periarticular connective tissue thickening is strongly correlated with duration of diabetes. The interphalangeal joints of the hands are always affected, but in more severe cases there is often multiple joint involvement. It has recently been associated with a short stature for age, and increased prevalence of retinopathy, nephropathy and thickening of the skin. This patient showed all of these features.

This patient manifests two disorders, one inherited and the other acquired; an incomplete form of Friedreich's ataxia and diabetic joint contracture. Both have been commonly associated with diabetes mellitus, but surprisingly, as far as we are aware, have not been described in the same individual. A number of possibilities might account for this. Firstly, the association of a characteristic limitation of joint mobility with long standing diabetes mellitus has been noted for less than 10 years and major studies of diabetes mellitus in patients with Friedreich's ataxia were conducted before this time so that early manifestations may have been overlooked. Secondly, because the ataxia nearly always antedated the onset of diabetes, patients usually succumbed to their progressive neurological disorder before the duration of diabetes was sufficient for limited joint mobility to become advanced and easily recognizable. The present patient is unusual in that her diabetes preceded her ataxia by 12 years, and also in that her neurological disorder has been non-progressive, allowing time for her advanced joint deformity to occur.

It may be that this case represents a series of random accidents rather than necessarily a systematic association. Its significance lies in illustrating the frequent association of diabetes with joint contractures, and the fact that in Friedreich's ataxia there is an increased incidence of diabetes.

SUMMARY

A case of juvenile diabetic joint contracture with Friedreich's ataxia is described in a 19-year-old girl. Review of the literature reveals no previous reports of the simultaneous occurrence of these disorders.

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