Spastic paraplegia associated with brachydactyly and cone shaped epiphyses

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SUMMARY Male uniovular twins presented at the age of 20 years with spastic paraplegia which had been slowly progressing over the years. Both have skeletal anomalies of their hands and feet with brachydactyly, cone shaped epiphyses, and an abnormal metaphyseal phalangeal pattern profile. In addition, they have a non-specific dysarthria and low-normal intellectual ability.

Hereditary or familial spastic paraplegia may be conveniently divided into the so-called pure type¹ and the syndromic group in which spastic paraplegia is associated with other features.² In the uncomplicated pure disease there is weakness and spasticity primarily involving the lower limbs, and with the exception of minimal sensory changes there are usually no signs in other body systems. This condition may be inherited in a variety of ways but dominant transmission appears to be the most common. The syndromic group has spastic paraplegia as a major feature but with other system involvement, for example, the skin, CNS, and optic tracts. They also show variable inheritance patterns. Although some skeletal abnormalities, for example, scoliosis, kyphosis, and pectus carinatum, have been reported in some of the recognised syndromes, there have been no reports of hand anomalies similar to those present in the twins in this report.

Family studies

Twin 2 and his wife were referred for genetic counselling on 2.7.85. An earlier pregnancy had ended in miscarriage and they had become concerned about the relevance of the husband's physical handicap for further pregnancies. He was known to suffer from a congenital spasticity of his lower limbs and had a similarly affected twin brother. The family are Punjabi Sikhs but the parents are non-consanguineous. The twins' father was 27 at the time of their conception and they have two healthy female sibs aged 25 and 20. Both parents are well and there is nothing contributory in the family history.

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TWIN 2

Twin 2 was born on 9.10.65 by forceps delivery 15 minutes after his brother. His birth weight was 3.5 kg and there was no note of any significant perinatal anoxia. By the age of 10 months he was pulling himself upright but was not walking by the age of two years. His head circumference at that time was recorded as 51.2 cm (above the mean but below the 98th centile for his age). By the age of four years he had developed lower limb spasticity with bilateral ankle clonus. He was walking without scissoring at the age of 11 years and a family photograph confirms that he and his brother were capable of playing football at that time. He remained within the normal school system and obtained a low grade in English at CSE level but was not graded in the other subjects.

On examination in the genetic clinic at the age of 20 years, his height was 163 cm (3rd centile), weight 46.9 kg (less than the 3rd centile), and head circumference 58 cm (98th centile). He had obvious severe, bilateral lower limb spasticity and stood with flexion of his trunk at the waist (fig 1a). He had some difficulty standing unsupported and walked with a marked scissor gait. Both lower limbs were thin and there was marked hyperreflexia of both legs with bilateral sustained ankle clonus and Babinski response. He had a pes planus deformity of both feet which were slightly externally rotated. The tips of all toes, particularly the big toes were broad and there was minimal soft tissue syndactyly between the second and third and to a lesser extent the third and fourth toes. There was some evidence of muscular weakness in his arms but there was no definite hyperreflexia. The tips of his thumbs were broad and there was striking clinical evidence of shortening of the fourth and fifth metacarpals of the hands (fig 2). There was some exaggeration of the normal abduction of the terminal phalanges of the first fingers of both hands and minimal soft tissue syndactyly of all fingers. He showed definite anterior bowing of his sternum but the spine was straight. His speech was slow, monotonous, and nasal and the palate was noted to be high. There was no other specific abnormality detected in the central nervous system. In January 1981 he was assessed by a

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educational psychologist and considered to be of low to average ability with limited language and comprehension. His personality was considered immature, withdrawn, and lacking in motivation at that time.

TWIN 1

Twin 1 was delivered by forceps and had a birth weight of 2.8 kg. There was no significant perinatal anoxia. He also was slow to walk and by the age of

FIG 1 (a) Twin 2 and (b) twin 1 showing truncal flexion associated with marked spasticity of lower limbs and pectus carinatum.

FIG 2 Clinically obvious shortening of fourth and fifth metacarpals.

four years had definite bilateral lower limb spasticity with sustained ankle clonus. At nine years he was noticed to be walking on his toes and had an Achilles tendon lengthening operation.

On examination at the genetic clinic in July 1985, his height was 164 cm (approximately 3rd centile), weight 43.3 kg (less than the 3rd centile), and head circumference 57 cm (midway between mean and the 98th centile). In physical appearance he is strikingly similar to his twin and has all the physical characteristics present and detailed in the second twin (fig 1b). An educational psychologist assessment confirmed him also to be of low to average ability with limited language and comprehension skills. He was educated within the ordinary school system but his attainments were low. His personality was thought to be more dominant than his twin brother.

Radiological findings

TWIN 2

X ray of the hands confirmed marked shortening of the metacarpals, particularly of the third, fourth, and fifth on the right and fourth and fifth on the left. The first metacarpal was also significantly shortened. The terminal phalanges of both thumbs were broad and short. There were cone shaped epiphyses of the proximal heads of the third and fourth second phalanges of both hands. The second middle phalanges bilaterally were also shortened. *X* ray of the feet confirmed shortening of the third, fourth, and fifth metatarsals bilaterally. There were cone shaped epiphyses of the proximal heads of the first phalanges of both feet (fig 3).

TWIN 1

X ray confirmed shortening of the metacarpals, particularly the fourth and fifth bilaterally and to lesser extent the first on the right. There were cone shaped epiphyses on the proximal heads of the second and fourth first phalanges. There was shortening of the third, fourth, and fifth metatarsals bilaterally with cone shaped epiphyses of both proximal heads of the first phalanges (fig 4). *X* ray of the chest confirmed a pectus carinatum deformity in both patients. No other skeletal abnormalities were noted.

Discussion

The hand anomalies are similar in both twins and the brachydactyly, presumably a consequence of the cone shaped epiphyses, is striking. The shortening of the metacarpals is confirmed by the metacarpophalangeal profile pattern³ and this technique was also of value in demonstrating the shortening of the terminal phalanges of the thumbs (fig 5). Although brachydactyly, particularly of the fourth metacarpal may be seen in a number of conditions, none of these appeared relevant to the patients in this report. Opjordsmoen and Nyberg-Hansen⁴ reported a number of families with spastic paraplegia associated with syndactyly and bladder disturbance. They made no reference to other hand anomalies such as brachydactyly and the twins do not admit to any bladder or sexual dysfunction. Pectus carinatum and dysarthria have been reported in other paraplegic syndromes apparently without other skeletal changes. The patients in this report presented with practically identical dysarthric patterns. Both

FIG 3 Radiographs confirming brachydactyly and cone shaped epiphyses in twin 2.

FIG 4 Radiographs confirming brachydactyly and cone shaped epiphyses in twin 1.

FIG 5 Metacarpophalangeal profile (Poznanski). Significant shortening of fourth and fifth metacarpals bilaterally in both twins. Shortening of third metacarpal in twin 2. Very short distal phalanges of thumbs bilaterally.

showed hypernasality with only slight pitch changes, leading to a monotonous tone, and both had a slow speed of utterance. Neither had other features of pseudobulbar palsy.

Blood group and HLA typing confirmed that the twins were uniovular. The marked similarity of the conditions in both patients and the absence of any other aetiological factor strongly suggests that the disorder is genetically determined. At present the mode of inheritance is not obvious, but single gene transmission would appear most likely.

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