Letters


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Familial hypoplasia of the thenar eminence: a report of three cases

Sir: Some individuals are born without certain muscles.1 These variations of the musculature are most often sporadic but hereditary absence of muscles has been reported.2-5 The pattern of inheritance in these families has been described as autosomal dominant. Congenital hypoplasia or absence of the thenar muscles is rare and only four sporadic cases have been reported.6-9 The purpose of this letter is to report three cases from two generations of a family presenting congenital hypoplasia of the thenar eminence. In addition two of these cases had no extensor pollicis longus muscles.

Case 1 was a 28-year-old man, referred to the department of neurology because of a left common peroneal nerve palsy which recovered during the following months. Since childhood bilateral hypoplasia of the thenar eminences had been noticed but apart from this there had never been any symptoms or signs of neuromuscular disease until the peroneal nerve palsy. A pronounced bilateral hypoplasia of the thenar eminence was found, with almost complete absence of the abductor pollicis brevis muscles (fig). In addition the extensor pollicis longus muscles were absent on both sides. Sensation, vibration and position sense of the hands was normal as was the rest of the neurological examination. Hand radiographs showed moderately hypoplastic scaphoid bones.

Case 2 was a 27-year-old woman, the sister of case 1, who since childhood had noticed hypoplasia of the thenar eminences. There had been no other symptoms or signs of

Fig Case 1, showing hypoplasia of both thenar eminences.
neuromuscular disease. On examination a pronounced hypoplasia of the thenar eminences was found. In addition the extensor pollicis longus muscles were absent. Sensation of the hands was normal, as was the rest of the neurological examination. Case 2 was married to a healthy man and had a daughter with normal hands.

Case 3 was a 66-year-old woman who was the mother of case 1 and 2. She had no musculoskeletal deformities. From all her life she had been aware of the hypoplasia of her right thenar eminence. Her brother, parents and husband were all known to have normal hands. From the age of 23 yr she had been suffering from rheumatoid arthritis. On examination the hands showed characteristic deformities due to rheumatoid arthritis. A moderate hypoplasia of the right thenar eminence was seen. The extensor pollicis longus muscle was present in this case. Sensation of the affected right hand was normal. Hand radiographs showed no congenital abnormalities, but the severe joint destruction of rheumatoid arthritis.

In two of these three cases the extensor pollicis longus muscles were also absent. The condition was without progression, and comprised only motor symptoms and signs. EMG of the abductor pollicis brevis muscles showed normal duration of the motor unit potentials and neither myopathic nor neuropathic abnormalities apart from a reduced number of motor unit potentials recruited at maximal effort. Sensory conduction studies of the distal part of the median nerve showed normal or low borderline values in all cases, and also the distal motor latency was normal. A significant reduction of the evoked motor response was found in two of the three cases. A differential diagnosis of distal muscular dystrophy or motor neuron disease was ruled out by the normal EMG findings (apart from a reduced interference pattern) and the clinical course. An entrapment syndrome of the median nerve at the wrist would comprise sensory symptoms and electrophysiological abnormalities would be expected. Only four sporadic cases of congenital hypoplasia or absence of the thenar muscles have previously been reported. In one of these cases also the hypothenar and pectoral muscles were hypoplastic. A neurophysiological examination was done in one case, showing normal EMG and nerve conduction values.

These conditions in all probability represent a primary failure of embryogenesis. The pattern of inheritance in the family reported was most likely autosomal dominant.

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