SHORT REPORT

"Pseudo" hypertrophic neuropathy of childhood

Masayuki Baba, Hiroto Takada, Hiroyuki Miura, Toshimi Okushima, Muneo Matsunaga

Abstract

A 9 year old boy had chronic progressive motor-sensory neuropathy that started in early infancy. He had enlarged nerves and pes cavus deformity. Motor conduction studies showed very dispersed, polyphasic compound muscle action potentials with conduction velocities around 2 m/s. A sural nerve biopsy showed severe loss of myelinated fibres. Two months of treatment with corticosteroids restored muscle power. During this time the enlarged nerves became normal and electrophysiological recovery was achieved. Chronically acquired neuropathy in infancy is strikingly similar to genetically determined neuropathy.

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It is often difficult to distinguish an acquired neuropathy from a genetically determined illness on the basis of the phenotypic presentation in an individual child. We present a 9 year old boy whose illness resembled genetically determined hypertrophic neuropathy. The condition, however, was reversed by corticosteroid treatment.

Case report

A boy with non-consanguineous parents began to walk at the age of 15 months, when the parents noticed that he had an awkward gait and that he often fell down. The walking

difficulty and hand clumsiness gradually worsened until the age of 9 when he visited us in a wheel chair. He complained of numbness in the hands.

On examination, swollen posterior auricular nerves were visible in the neck. The limb muscles were severely weakened and thin distally. The intrinsic hand muscles were totally paralysed. His feet and toes were almost immovable. There was no fasciculation in the limb muscles. Pes cavus deformity was noted bilaterally. He could not stand without support. Sensory loss in all modalities was seen distally. Tendon reflexes were absent. Firm and thickened nerves were palpable around the elbows and the knees. Sphincter function was normal. There was no Babinski sign. He had normal intelligence.

Routine laboratory tests, including tests for antiganglioside antibodies, were all normal or negative. Protein concentration in CSF was 68 mg/dl with no pleocytosis. Brain and spinal MRI showed no abnormalities.

Massive oedematous swelling of the sural nerve was noted during biopsy: microscopically, there was endoneurial oedema and myelinated fibres were almost completely lost. There were some cellular infiltrates in the subperineurial space. Electron microscopy showed only occasional onion bulbs.

High dose intravenous methylprednisolone (25 mg/kg/day) was given for three days, followed by oral prednisolone (2 mg/kg/day). A week after the treatment started, numbness in his hands diminished, distal superficial sensation recovered gradually, and his muscles became stronger. During a four week period his walking ability was restored, the nerve swellings subsided, and the posterior auricular nerves became invisible. Protein concentration in CSF was reduced to 36 mg/dl. During two months of treatment, grip power increased to 25 kg. He was then discharged able to walk and started to enjoy normal school life.

Electrophysiology

Motor and sensory nerve conduction studies were performed with standard techniques and percutaneous stimulation. Skin temperature was above 34°C during the procedures. Electromyography was carried out with a concentric needle electrode.

Department of Neurology, Institute of Neurological Diseases, Hirosaki University School of Medicine, Hirosaki 036, Japan

M Baba
H Takada
H Miura
T Okushima
M Matsunaga

Correspondence to:
Dr Masayuki Baba,
Department of Neurology,
Institute of Neurological
Diseases, Hirosaki
University School of
Medicine, Zaifu-cho 5,
Hirosaki 036, Japan.

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Summary of nerve conduction studies

	DL (ms)	CMAP-Amp (W/E) (mV)	Dur (W/E) (ms)	MCV (m/s)
Before steroids:				
Right median	53.6	0.13/0.08	6/8	1.7
Left median	34.6	0.21/0.18	35/60	2.7
Right ulnar	15.8	0.36/0.32	49/78	1.8
Left ulnar	10.2	0.32/0.30	85/115	2.2
Two months after steroids:				
Right median	8.2	0.36/0.24	25/33	7.3
Left median	8.1	0.84/0.55	48/119	16.2
Right ulnar	6.1	2.0/1.4	24/59	11.4
Left ulnar	5.8	2.4/1.1	47/113	14.8

DL = Distal latency; CMAP = compound muscle action potential; Amp = amplitude; W = wrist stimulation; E = elbow stimulation; Dur = duration; MCV = motor conduction velocity.

Left median nerve
conduction study.
W = wrist stimulation;
E = elbow stimulation.

Results

The table summarises the nerve conduction results before and two months after steroid treatment. Before treatment extremely dispersed compound muscle action potentials (CMAPs) with amplitudes of 0.13 to 0.36 mV were obtained from the hand muscles (figure). Conduction velocities were between 1.7 and 2.7 m/s, and distal latencies were between 10.2 ms and 53.6 ms. No CMAPs could be elicited from the foot muscles. Electromyography of the wrist extensors and the tibialis anterior muscle showed some polyphasic motor unit potentials and occasional fibrillation potentials. Sensory potentials and somatosensory evoked potentials could not be elicited.

During corticosteroid treatment, improvement of latencies and amplitude of CMAPs was remarkable. The CMAP amplitude after distal stimulation increased by more than 500% in the left ulnar nerve, and distal latency time of the right median nerve was shortened to 8.2 ms from a pretreatment value of 53.6 ms. The CMAPs of both the median and ulnar nerves became compact and bigger. As well as the main deflections, some isolated late units, not seen before treatment, were elicited. During the two months of treatment MCVs increased to 7 to 16 m/s. Continued treatment with prednisolone (30 mg on alternative days) maintained the improvement in nerve conduction.

His parents, grandparents, and two siblings had no signs of neuropathy and had normal nerve conduction values.

Discussion

The clinical features of this child, together with raised CSF protein concentrations and extremely slow conduction velocities of around 2 m/s, satisfy the diagnostic criteria for hereditary motor-sensory neuropathy type III, or Dejerine-Sottas disease.¹ Few onion bulbs, however, was a finding against it, and small foci of cellular infiltrates and conspicuous intrafascicular oedema strongly suggested an inflammatory origin^{2,3} or chronic inflammatory demyelinating polyneuropathy (CIDP). Motor conduction studies failed to show a conduction block before treatment; nevertheless, a combination of extremely slow motor conduction velocities and disproportionately short distal latencies found in the ulnar nerves suggested the presence of multifocality, one of the essential features of acquired demyelinating neuropathies.⁴

Changes in nerve conduction after the treatment were remarkable in this patient. During steroid treatment, amplitude of the left ulnar CMAP became nearly eight-fold greater than before the treatment; decrease in latency time and synchronisation of CMAPs simultaneously developed. Some newly recruited late units were also identified. Rapid increase in muscle power is another sign of recovery in impulse transmission along revived motor fibres after demyelination.

Steroid responsive neuropathy in childhood was first mentioned by Byers and Taft.⁵ Recently, Sladky *et al* mentioned nerve enlargement in their cases with childhood CIDP.⁴ The present case is unique, because the conspicuous nerve thickenings subsided within two months of treatment. It seems, therefore, that the thickened nerves were not really hypertrophic; massive oedema in and around the nerve was the probable cause of the nerve swellings in this patient.

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