Painful cauda equina schwannoma simulating Charcot-Marie-Tooth disease

Sir: Pain is almost always the principal symptom of patients who have tumours of the cauda equina. Painless tumours at this site, although rare, do occur and should be considered in the differential diagnosis of pain free patients presenting with progressive weakness in the lower limbs. The following case report demonstrates this point.

A man, age 38 years, was admitted with 10 years' history of progressive weakness of the legs, the left being more severely affected. There was no history of any pain or paresthesia in his back or legs. Other symptoms included a recent constant feeling of urgency, occasional nocturia, constipation, and weak penile erection. Previously he was admitted to another hospital because of a left ankle sprain, leg weakness, and a gait disturbance, at which time a diagnosis of peroneal muscular atrophy was made. The abnormal neurological signs were confined to the lower limbs. Moderate to severe weakness and wasting, especially of hip extensors, foot dorsiflexors, extensors of toes, foot extensor and invertors, were present with bilateral foot drop. Knee jerks, ankle jerks, and plantar responses were normal. There were no sensory changes. The rectal sphincter was hypertonic. Straight leg raising to 90° produced no pain or discomfort. Lumbar spine radiographs were normal. Nerve conduction studies with standard techniques showed normal maximum conduction velocities in right median, ulnar, tibial, and sural nerves, and borderline conduction velocities in left tibial nerve. Supramaximal percutaneous stimulation of the peroneal nerves produced no muscle action potential. Needle electromyography showed extensive denervation potentials in glutei maximus and in the muscles innervated by both sciatic nerves but no abnormality in paraspinal muscles. Myelography revealed a mass at the cauda equina region. Spinal fluid protein was elevated (1-28 g/l). Urodynamic studies were consistent with a spastic neurogenic bladder. Laminectomy of D12 and L1 vertebrae was performed and a large Schwannoma was removed totally.

Cauda equina tumours are rare. Occasional cases present with unusual clinical fashions such as pseudoclaudication, foot ulceration, subarachnoid haemorrhage, papilloedema, and sensory ataxia, but it is generally accepted that pain in the back and lower limbs is nearly always the most important and early presenting symptom. Pain may be of sudden or gradual onset, usually tends to become constant, and characteristically worsens at night. Allen and Spiller reviewed the literature of cauda equina tumours and both found only one case of cauda equina compression with no pain, reported by Volhard. Campbell could find clinical details of only two cases of painless tumour of the cauda equina and presented one of his own. In a series of 20 patients from the National Hospital, Queen Square, only one had no pain and presented with left leg weakness for seven years; but finally he developed severe lumbar pain. In an Oxford series of 70 patients, six complained of painless progressive weakness of legs; only three had bilateral leg weakness, and pain was the presenting symptom in 31 out of 34 patients with neurofibromas of the lumbar-sacral region.

In sum, a case is reported of a patient with a Schwannoma of the cauda equina who carried the diagnosis of Charcot-Marie-Tooth for 10 years. There was slowly progressive motor dysfunction without pain or sensory loss during this time. Electromyography excluded Charcot-Marie-Tooth disease, and indicated a lesion of the cauda equina and, therefore, mandated myelography. This emphasises the potential diagnostic significance of proper electromyographic study in patients with progressive neuromuscular disease.

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References


Letters

Use and misuse of the Roussy-Levy eponym

Sir: On a clinical and electrophysiological basis only, Yudell, Dyck and Lambert in 1965 described a family with a dominantly inherited form of peroneal muscular atrophy which they called Charcot-Marie-Tooth. As this group of patients showed substantially reduced motor conduction velocity they were assumed to have onion bulbs in their peripheral nerves and were therefore labelled hypertonc Charcot-Marie-Tooth. Four of the nine patients in this kindred had a disorder of movement similar to essential (familiar) tremor of unusually great amplitude. This association of signs was considered to be an example of the Roussy-Levy disease. The use of this eponym for such patients seems to us to be illogical since in 1906 P Marie described a family with a dominantly inherited peroneal muscular atrophy, hypertrophic nerves and essential tremor of great amplitude. Boveri in 1910 provided the post mortem material of the member of the kindship who had the tremor of the largest amplitude. Since that time this type of hypertrophic neuritis has been known as the Pierre Marie-Boveri type. P Marie did not say that his patient had the hypertrophic variety of Charcot-Marie-Tooth disease associated with essential tremor and
stressed that the family he had reported, because of the presence of tremor, was different from the hypertrophic neuritis described by Dejerine and Sottas. Thus an original and sound description of a neurological entity was made from accurate clinical and pathological data.

Roussy and Levy in 1926 also described a family with a dominantly inherited disease featuring pes cavus, absent tendon jerks and difficulty in standing and walking. They emphasised that their patients did not have a cerebellar deficit and that the disease was probably non-progressive. They did not realise that, as emphasised by Symonds and Shaw in 1926, they had described a "forme fruste" of Charcot-Marie-Tooth disease. Furthermore although four of their seven original patients had essential tremor of very mild amplitude they did not emphasise the existence of essential tremor as a trait of the disease. Lapresle in 1956 re-examined four of the original patients of Roussy and Levy and noted that two of them had the bodily appearance of Charcot-Marie-Tooth disease. In the original case IV Lapresle and Salisachs noted that essential tremor of a very low amplitude had appeared and that motor conduction velocity was very reduced. That patient had onion bulbs in the sural nerves. Two other cases of the kinship originally described by Roussy and Levy have shown similar electrophysiological and pathological features. Salisachs and Lapresle pointed out that the entity described by P Marie and Boveri was probably only different in degree from that reported by Roussy and Levy. This idea was later acknowledged by Dyck.

What eponymous title would the writers following the authors from the Mayo Clinic give to the numerous patients with Charcot-Marie-Tooth disease, essential tremor and normal or slightly reduced motor conduction velocity? It should be noted that Professor Raymond in his lessons at La Salpetriere in 1900–1901 first reported a brother and a sister with Charcot-Marie-Tooth disease associated with essential tremor. As the nerves of these patients were enlarged he assumed that they had onion bulbs and furthermore he did not accept any difference between Charcot-Marie-Tooth disease and the hypertrophic neuritis of Dejerine-Sottas. He may be criticised for this later idea but he categorised his patients as Charcot-Marie-Tooth disease associated with essential tremor and hypertrophic nerves. Therefore Raymond seems to have been the first to draw attention to this association although he has never been given the benefit of eponymous honours.

Further difficulties arise with the use of the eponym Roussy-Levy. Some authors apply it to the association of pes cavus, ataxia and areflexia which is not only incorrect (for the patients reported by Roussy and Levy did not have such signs in 1926, 1956 or when one of the present authors (PS) examined three of them in the seventies) but also imprecise. Indeed, such association can be seen in Charcot-Marie-Tooth disease (whatever the motor conduction velocity values), Friedreich's ataxia, other spinocerebellar degenerations, Refsum's disease etc.

Since the first description by Roussy and Levy some have thought that this disorder was a "forme fruste" of Charcot-Marie-Tooth disease or an "abortive type" of Friedreich's ataxia, whilst still others considered it as a transition form between the two. This linkage between Friedreich's ataxia and Charcot-Marie-Tooth disease was sustained by misusing the reports of kinships in which Friedreich's ataxia, Charcot-Marie-Tooth disease and Roussy-Levy disease were incorrectly said to occur together and also by the existence of families in which some members would show signs of both Friedreich's ataxia and Charcot-Marie-Tooth disease. Such views are no longer tenable because Charcot-Marie-Tooth disease share many clinical features with Friedreich's ataxia and has been shown to mimic it closely. Indeed, club foot, absent tendon jerks in the limbs, errors in vibratory sense, two point discrimination and position sense, nystagmus, a positive Romberg sign, kyphoscoliosis, extensor plantar responses, dysarthria and inco-ordination (mimicking cerebellar disease) can be present in Charcot-Marie-Tooth disease. In such cases this latter disorder masquerades as Friedreich's ataxia.

Patients presenting signs of two different disorders cannot be assumed to represent transition forms between these two disorders as demonstrated by the fact that in the largest series of patients with Charcot-Marie-Tooth disease and Friedreich's ataxia studied with modern techniques no kinship was found to have both disorders, transition forms or any other spinocerebellar degeneration.

Modern textbooks sometimes suggest incorrectly that there are transition forms between Friedreich's ataxia and Charcot-Marie-Tooth disease. Other authors feel that the Roussy-Levy eponym describes patients with Friedreich's ataxia in whom peroneal muscular atrophy is prominent. In addition some families have been wrongly considered to have some members with Friedreich's ataxia while other affected members had other spinocerebellar degeneration (both with and without distal weakness and wasting). These views may mislead some readers into believing that there is a continuum between Friedreich's ataxia, Charcot-Marie-Tooth disease and other spinocerebellar degeneration both with, or without, distal weakness and wasting (see figure).

Fig. Arrows represent proven links and discontinuous lines represent unacceptable transitions.

In conclusion the use of the eponym Roussy-Levy seems to us to be historically misleading, unhelpful and may perpetuate conceptual inaccuracies concerning some heredodegenerative disorders.
Matters arising

Histocompatibility antigens on astrocytoma cells

SIR: We have read with interest the article on histocompatibility antigens on astrocytoma cells by Hirschberg et al. recently published in your journal. The authors state that dissociated cells from astrocytommas of various degrees of malignancy do not express HLA-D/DR determinants. We recently demonstrated that monoclonal anti-HLA-DR antibodies reacted with glioma cells from several established lines, as shown in an antibody-binding radioimmunoassay. Monoclonal anti-HLA-DR antibodies were shown to lyse specifically 51Cr-labelled glioma cells in the presence of complement. Absorption of anti-HLA-DR antibodies by glioma cells abolished their cytotoxicity against blasts isolated from a common acute lymphoblastic leukaemia (c-ALL) line. Immuno-precipitation of solubilised 125I-labelled membrane proteins from glioma cells by monoclonal anti-HLA-DR antibodies revealed two polypeptide chains of 28 and 33 kDa, characteristic of HLA-DR antigens. It must be stressed, however, that not all glioma cell lines tested expressed HLA-DR antigens (three of eight tested). Hirschberg et al. point out that cell lines derived from solid tumours are probably not as suitable as primary tumour cell cultures in the study of representative antigenic determinants on the cell surface. This is true, but primary cultures are already a selection of the original cells. We have

References


Training for clinical practice

SIR: The editorial commentaries by Dr Hopkins and Professor Marsden (December, 1981) raise issues of relevance to clinical neurosciences education. Dr Hopkins records that established clinical training is founded on specific nosological entities, which his table shows to be uncommon in practice. Moreover, as he and Dr Fitzpatrick report in their accompanying article, patients present to neurologists with complaints of disordered form and function. Professor Marsden concludes from Dr Hopkins’ figures that the majority of common neurological problems in the United Kingdom will be managed by primary care physicians; it is important to know, therefore, whether current training equips practitioners with the requisite proficiencies to execute these responsibilities.

As part of a study to define educational objectives in undergraduate neurosciences and to establish competencies in clinical neurology relevant to future activities...
Use and misuse of the Roussy-Levy eponym.

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*J Neurol Neurosurg Psychiatry* 1982 45: 938-940
doi: 10.1136/jnnp.45.10.938-a

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