only a mild degree of nerve fibre loss. The corpus callosum showed severe loss of nerve fibres, increased numbers of glial cells, reactive astrocytes and focal necrosis. The caudate nucleus and internal capsule on the left side were normal. Only mild neuronal loss was seen in the left globus pallidus. The cerebellum and the whole right hemisphere were normal. Scattered giant axonal spheroids were present in the posterior portion of the rostral medulla. No evidence of glioma was found. The abnormalities described were diagnostic of a widespread leukoencephalopathy due to radiation and/or chemotherapy.

Most extraneural metastases from gliomas occur after craniotomy although at least eight cases\(^1\) have been reported, in the absence of surgery. The diagnosis of systemically metastatic central nervous system tumours is not easy to make and is probably not often considered. Weiss\(^4\) established four criteria: first, the presence of a single tumour which is histologically characteristic of a primary glioma; second, a clinical history indicating that initial symptoms were due to this tumour; third, a complete necropsy excluding the possibility of any other primary site; and fourth the identical morphology of the CNS tumour and the distant metastases with due allowance for differences in degree of anaplasia. The use of glial fibrillary acidic protein, a cytoplasmatic marker for identifying glial cells,\(^5\) provides a more definitive histological characterisation of tumours of neural origin and allows for the diagnosis of metastatic glioma in the absence of the classical criteria.\(^6\)

The present case is unique in that three rare events in the treatment of a glioblastoma occurred in concert, namely systemic metastasis, cure of the glioma at the primary site and treatment induced leukoencephalopathy. Death was from complications secondary to bone marrow infiltration by glioma cells. The patient developed progressive neurological dysfunction from white matter degeneration following radiation and chemotherapy while the primary tumour appeared to have been eradicated. The major problem during the patient’s last several months related to his pancytopenia. The diagnosis of metastatic glioma was not considered until necropsy. Marrow suppression was initially thought due to chemotherapy. Terminally, when a bone marrow biopsy produced malignant cells, a second malignancy was thought to have developed. This case suggests that combined radiation and chemotherapy were effective in treating the glioma but were the cause of white matter degeneration. As prolonged survival becomes more common with this tumour, clinicians may see an increasing number of similar complications. Staining for glial fibrillary acidic protein may preclude a lengthy and costly search for a second primary malignancy.

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Optico-acoustic atrophy in distal spinal muscular atrophy

Sir: The triad of hereditary motor and sensory neuropathy, optic atrophy and deafness was first reported in a kindred of two brothers and their nephew by Rosenburg and Chutorian in 1967.\(^1\) A similar clinical syndrome in a brother and sister was described in 1970.\(^2\) But for the sensory manifestations, the clinical features of hereditary motor and sensory neuropathy might be confused with the distal form of spinal muscular atrophy. We report a case of distal spinal muscular atrophy associated with optico-acoustic atrophy and believe that this association has not been previously described.

The patient presented at the age of 10 years with visual impairment. Three years later she became deaf and was shown to have severe bilateral sensori-neural deafness 12 years later. At this time she also complained of clumsiness of her hands and was found to have distal muscle wasting and weakness in both upper limbs. There were no sensory signs and the deep tendon reflexes were retained.

Fifty-one years after initial presentation, she was unable to hear and understand speech. Visual acuity (aided) was 6/60, N18 bilaterally. The symptoms and signs in the upper limbs remained unchanged. There was no family history of neuro-muscular disease, blindness or deafness and she had had no children.

Examination revealed bilateral optic atrophy with no pigmentary retinopathy. There was marked wasting and weakness of the small muscles of the hands and forearms with preserved reflexes. There were no sensory signs and no thickened peripheral nerves.

Glucose tolerance test, serum thyroxine and creatine kinase were normal. No acanthocytes were seen in the peripheral blood. Cervical spine radiographs showed mild cervical spondylosis. CSF protein and gamma globulin levels were normal. Nerve conduction studies showed normal motor and sensory conduction velocities in upper and lower limbs. Sensory action potentials were of normal amplitude (right ulnar 10\(\mu\)V, right sural 12\(\mu\)V). Electromyography showed evidence of chronic partial denervation with polyphasic potentials and giant units to 12\(\mu\)V in proximal and distal muscles in upper and lower limbs. Spontaneous activity was sparse. Muscle biopsy (left deltoid) showed fibre-type grouping compatible with the electromyographic findings. Visual evoked potentials (checkboard pattern reversal) were delayed bilaterally. The

References

electroretinogram was normal. Brain stem auditory evoked potentials were poorly formed but of normal latency. Audiometry confirmed severe sensory neural deafness and a CT scan was normal.

The association of hereditary motor and sensory neuropathy with optic atrophy was first reported in 1889.3 and a number of other families in which both disorders occur have since been described.4–6 Likewise there is an association between hereditary motor and sensory neuropathy and deafness. Of a series of 225 patients with hereditary motor and sensory neuropathy, four were found with sensori-neural deafness.7 Rosenberg and Chutorian reported a family in which three members had optico-acoustic atrophy and hereditary motor and sensory neuropathy.1 These patients had moderate slowing of the motor conduction velocities without thinned peripheral nerves. They discussed the relationship between the hereditary spino-cerebellar degenerations, optic atrophy, nerve deafness, hereditary motor and sensory neuropathy and hereditary sensory neuropathy and concluded that a considerable overlap between these conditions exists. Iwashita et al, described a pair of siblings with the same triad of conditions.8 One of these also had evidence of sensory involvement. These cases had normal motor conduction velocities and evidence of widespread denervation. The amplitude of the sensory action potentials was not reported in either of these patients. Both groups note the superficial similarity of these conditions to Refsum's disease but consider that the differences are more fundamental, with no increase in serum phytic acid or CSF protein, no retinitis pigmentosa, anosmia, ichthyosis or ECG abnormalities.

Our case is similar in many respects to those mentioned above. The onset of symptoms in the present case is, however, slightly later than in those previously reported. The progression of the disease in our case appears to have arrested and to have been stable for the past 30 years or so. No long term follow-up data is available on the other cases for comparison.

Although the electrophysiological data are not described in detail, there is clinical evidence of sensory involvement in the previously reported families. Our patient had no clinical or electrophysiological evidence of sensory neuropathy thus favouring a diagnosis of distal spinal muscular atrophy.

This case suggests that optic atrophy and deafness are not only an occasional feature in hereditary motor and sensory neuropathy, but also a rare finding in the distal form of spinal muscular atrophy.

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References


A case of tabes dorsalis with tonic pupils and lightning pains relieved by sodium valproate

Sir: We report the case of a woman who had had tabes dorsalis accompanied by lightning pains for 30 years. The pains were relieved within a few weeks by sodium valproate, but an attempt to replace this drug by carbamazepine was unsuccessful. The patient also has tonic (Holmes- Adie) pupils. The patient is a married woman born in 1914 who was first seen in August 1982, when aged 68 years, at the Centre for Pain Relief, Mersey Department of Medical & Surgical Neurology, Walton Hospital, Liverpool. She complained of “aching legs” and lighting girdle pains in the chest which she had been having for 30 years. The pains came in bouts lasting two or three weeks; she experienced four or five bad bouts and two or three less bad attacks per annum. The pains were exacerbated by cold, “tension”, and anger; and ameliorated by warmth (hot water bottle) and rest. She experienced “severe tingling” on sponging herself.

She had acquired a treponemal infection from her husband at the end of the war, during which he had served in the Merchant Navy. He was reported in 1983 as having serology consistent with having had a treponemal infection in the past; he died in October 1985 of renal and cardiac failure complicating carcinoma of the bladder. The patient herself was first seen at the Special Clinic in the Liverpool Royal Infirmary in 1958, when tabes was diagnosed. Irregular pupils, described as Argyll-Robertson, were reported at that time. She was treated with penicillin, and found to be Wassermann-negative in 1960 (blood and CSF). Further serological examinations in 1980 and 1984 confirmed only a past infection. The patient has a family history of hypertension, and is herself hypertensive, her blood pressure having been as high as 200/120 mm Hg; this is being treated with propranolol 40 mg/day. She has also been investigated for peptic ulcer, as she has a long-standing (and familial) history of heartburn. She has bilateral cataracts of long standing. Only one of her five siblings is still alive. The patient has a son and daughter, both born before the Second World War. Both are healthy and married, and each has two healthy children.

At presentation in 1982, the patient underlined 15 words on the McGill Pain Questionnaire. Conventional analysis1 showed that 90% of word groups in the somatic category were chosen, 100% in the evaluative category, 100% in the miscellaneous category, but only 25% in the affective category. The (rank) intensity score was 45, or 75%. Overall, the picture was one of severe organic pain with remarkably little functional overlay, in sharp contrast to the score patterns obtained in cancer2 or backache.3 When asked which pain descriptors she would most like to be rid of, she unhesitatingly replied “stabbing and scalding”.

On examination (which was during a bout, but not during an attack, of pain) she was 150 cm tall and weighed 55–2 kg. The painful area was symmetrically distributed in the T5 segments; the right upper thorax and both legs exhibited allodynia. Tone and power were normal for her age, but tendon reflexes were universally absent, and no superficial reflexes could be elicited. She did not exhibit the slowed withdrawal times from noxious stimulation which are usually associated with tabes dorsalis. Sensation of passive movement was normal and symmetrical at all joints tested. Pinprick sensa-
Optico-acoustic atrophy in distal spinal muscular atrophy.

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