A predominantly cervical form of spinal muscular atrophy

We have reservations about the paper by Dr Goutieres et al., though not the concept itself. Documenting the clinical and histological findings in five infants, they describe a condition "A predominantly cervical form of spinal muscular atrophy". Cervical spinal muscular atrophy (SMA) is a real entity and has been reported previously. It is probably due to a disturbance in the lower cervical segments of the spinal cord resulting in muscle wasting and contractures of the upper extremities and normal lower limbs.

They have not mentioned any necropsy findings. They need other sources of confirmation since they attempt to describe a newly defined clinical condition. In atypical posterior spinal muscular atrophy (PSMA), described by the authors should have ruled out a possible cervical hydromyelia or syringomyelia with amyotrophy or even a congenital cervical spinal tumour, and familial syringomyelia is well-known. For this purpose a myelogram—CT or an MRI of the cervical spine should have been done. Imaging spinal areas by MRI has almost revolutionised our concepts. We have seen two children with cervical spinal pathologies, one with a dermoid tumour and one with suspected syringomyelia (unoperated upon as yet, so no tissue diagnosis is available). Both children had symptoms mimicking SMA. In each case, MRI was the greatest help and was the most straightforward method. In conditions running a more benign course such as monomeric amyotrophy, studies with H2 or metrazamide CT have demonstrated atrophy of the related spinal cord.

Muscle biopsy findings in the cases of Dr Goutieres et al. were all consistent with "neurogenic fascicular atrophy". In the absence of giant type fibres which are typical for SMA in early infancy, muscle biopsy revealed the presence of denervation, the extent of regeneration and the chronology of the process, but did not conclusively disclose the precise anatomical localisation of the denervating event. There is no comment on whether there were any giant type 1 fibres in the muscle biopsies.

Defining prognosis in medical coma

I appreciated David Bate's well written editorial on the process and limitations of establishing prognoses in patients with medical coma. Since we collaborated on the international non-traumatic coma study, perhaps I may make one or two points not strongly emphasised by Dr Bates.

The editorial generally summarises accurately the international study's results but omits emphasising a dimension which may be critical to self-evaluation. That is, the study excluded all patients whose coma did not result either from known organ failure, or from known exogenous causes such as a deprivation of oxygen supply or an excess insulin dose. All cases with self-induced coma-causing drug poisoning as well as all cases in which aetiological diagnosis was uncertain were automatically excluded. The reasons are straightforward: nearly all such patients survive intact with intensive care including some with a flat EEG and fixed pupils lasting for a day or more.

I regret that I disagree with Dr Bates in his contention that since only a small number of several thousand patients can reduce the theoretical error of 5% in predicting poor outcome, one cannot make decisions based on unfavourable early signs, however bad they may be. What about the next 1000 patients if they survive to dementia? Most Americans are aware of the meaning of probability odds. Given 20:1 odds they ought to be sitting in bed or in a vegetative state or no longer have a reason to live, but they certainly would not do so if they risked having to witness and indefinitely support a pained and crippled being if that was the cost of losing the bet. Dr Bates does not mention the only damaging downside statistical feature anywhere in his editorial. Many Americans are becoming increasingly apprehensive about being rescued from an early death by critical care measures only to face lives permanently blighted by intractable pain, severe physical disability, cognitive impairment or some combination of all three. When we advise patients or their families on day 1, 3, 5 or later that if they continue to receive maximal care, they or their loved ones may have a 2% or 5% statistical chance of a good recovery we also tell them that continued survival also includes at least 50-64% of patients associated with permanent severe disability. Facing such choices, a few will say, "Please do everything, doctor". In my own experience, however, most will urge, "please be merciful—he/she couldn’t stand living as a permanent cripple, much less being a hopeless burden on the family".

The humane decision of who and when to treat and how long necessarily delicate, difficult and sometimes painful for the physician; it is an even greater burden for the family. At least by the US Constitution, the doctor is neither the only party nor the major decision in this situation, the patient is. Evidence in this country, is that we physicians are under-fulfilling our responsibility on this critical matter.

In my year, Derek Humphrey was the author of a small monograph entitled "Final Exit", which offers direct advice on how to commit suicide for those who, for whatever reason, are considering it. It reached the national bestseller list. The accompanying New York Times news story included comments from bookellers, journalists and potential patients that implied that physicians often overstate or unduly