

CARPHOLOGY by A Fo Ben



Pract Neurol 2011; 11: 56

CMT spelled out

The race to sequence the whole genome for neurological disorders has started. An individual from a Charcot–Marie–Tooth (CMT) family had his entire genome detailed, identifying a potential mutation in *SH3TC2* (the SH3 domain and tetratricopeptide repeats 2 gene). Genotyping the family demonstrated that those individuals with a mutation in both copies of *SH3TC2* had the CMT phenotype. Interestingly, those who were heterogenous for *SH3TC2* showed a greater risk of mononeuropathies, such as carpal tunnel syndrome. This is an eloquent description of how state of the art research genetics can provide clinically relevant results.

N Engl J Med 2010;**362**:1181–91.

The plural of anecdote is not data

Learning from anecdotes—case by case—is a very slow process. A Fo Ben learned that charging people with multiple sclerosis £10 000 for injections of macerated bovine brain and spinal cord tissue can get you struck off the medical register. Dr Robert Trossel advertised his stem cell therapy without warning about the risks of prion infection; indeed, the stem cells were never intended for human use. The GMC said that he had exaggerated the benefits of treatment based on 'anecdotal and aspirational information'.

<http://www.bbc.co.uk/news/health-11425435>.

Dr Plum: champion of consciousness

Dr Fred Plum was a 'giant of neurology' who will be sorely missed following his death last summer. He was inspired to study poliomyelitis following the death of his sister. His expertise in this area led to him forming a respiratory centre in Seattle. Via his study

of resuscitation of people who had overdosed with barbiturates, he became a leading expert in coma, coining the term 'locked in syndrome' and (with Bryan Jennett) 'persistent vegetative state'. That his death was from primary progressive aphasia was particularly unfair, as it will be for his bedside teaching, published papers and communication that he will be remembered.

Channel No 1

It must have been the month for genetic studies—a dominant negative mutation in the *TRESK* potassium channel has been implicated in a pedigree of familial migraine with aura. *TRESK* is expressed in the spinal cord where it modulates cortical excitability, and has previously been implicated in pain processing and cortical spreading depression. A frameshift mutation in the gene coding for *TRESK*, which renders the channel non-functional, was found in all members of the extended family who had migraine, and in none of those family members who did not. This identification of a candidate gene both provides a putative mechanism and a potential therapeutic target for these patients. Investigation of other affected families is certainly warranted.

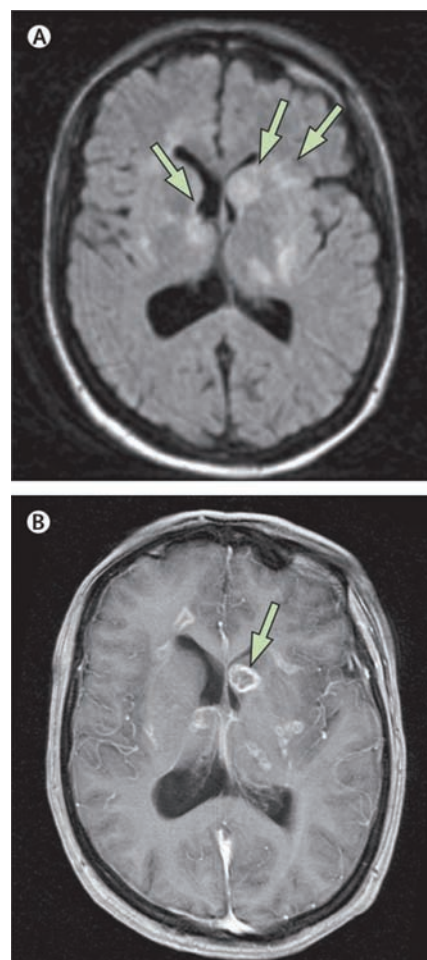
Nat Med doi:10.1038/nm.2216.

Once bitten...

A recent case report has highlighted both the more esoteric dangers of horse riding and the importance of taking a detailed history. A 37-year-old woman presented with a 3 day history of fever, headache and confusion. CSF and blood cultures grew Lancefield group C streptococci. On questioning, her relatives disclosed that she was an enthusiastic horsewoman, and had suffered what had appeared to be a minor bite from a healthy horse a week previously. Treatment with benzylpenicillin continued for 8 weeks for *Streptococcus equi*

meningitis, with significant clinical improvement, although the patient remains severely amnesic (see axial MRI images below).

Lancet 2010;**376**:1194 (figure used with permission).



(A) Multiple lesions with increased signal intensity on FLAIR images. (B) Ring shaped gadolinium enhancement.

A Fo Ben is always on the lookout for suitable Carphology titbits and comments on what has been included. Email the editor-in-chief if you come across anything.