

Highlights from this issue

Phil E M Smith, Geraint N Fuller

Neurology is a diagnostic specialty. While it is true that neurologists are spending more time managing neurological disorders than they ever did—in light of the range and complexity of treatments available—the management and prognosis still all stem from the diagnosis.

However, not all diagnoses are equal and we have several cases that illustrate this. Richard Ibitoye and colleagues (*see page 496*) describe a patient with epilepsy and ovarian failure (clinical diagnosis) who also had a leukodystrophy (radiological diagnosis), in turn related to a mutation in *EIF2B5* gene (genetic diagnosis). Matthew Evans and colleagues explore a patient with the ‘16 syndrome’, an educational anatomical diagnosis (*see page 484*); Mihaela Boca and colleagues (*see page 458*) discuss diagnosis that evolved from clinical to radiological to pathological and aetiological, in a young man with basal ganglia necrosis, describing their iterative diagnostic process as the ‘best fit’ approach.

Simon Rinaldi and colleagues (*see page 488*) report a patient with recurrent back pain but without weakness or sensory loss and argue this is the relapsing form of chronic inflammatory demyelinating polyradiculoneuropathy (CIDP). CIDP is a syndromic diagnosis and they suggest that their patient's illness, even though not fitting within the previously described syndrome, still reflects the same pathological process and thus broadens the range of clinical phenotypes associated with this pathology.

Most syndromic diagnoses are based on an appropriate clinical phenotype along with supportive investigations that exclude relevant alternatives—think about making a diagnosis of Guillain-Barré syndrome in a patient with ascending weakness

and cytoalbuminologic dissociation in cerebrospinal fluid. GF (conflict of interest alert) argues in the ‘mimics and chameleons’ article (*see page 439*) that we should diagnose ‘Bell’s palsy syndrome’ when a patient presents with the characteristic isolated unilateral facial weakness that comes on over days, subject to a few exclusions, rather than consider ‘Bell’s palsy’ simply as a facial nerve weakness for which no cause is found.

One emerging clinical syndrome is the ‘postural orthostatic tachycardia syndrome’ (POTS). Pearl Jones and colleagues (*see page 431*) bring us up to date with this syndrome, what is known and what is not. Despite being only relatively recently described, we have several ways to help patients with POTS; that is, if the diagnosis has been made.

Some clinical syndromes are the starting point for the diagnostic process. Ellie Marsh and Maria Cauci present a patient with a dropped head and discuss the differential diagnosis and diagnostic strategies for patients with this uncommon but distressing problem (*see page 445*).

The neuromyothology series continues to save you time: Jonathan Schott and Martin Rossor argue that the palmomental reflex can now be retired from clinical assessment (*see*

page 500). We have included Marty Samuels' succinct referee commentary on the paper, in which he recognises the inevitable continued role for this sign as part of the flamboyance of 'roundsmanship'.

You have the opportunity to try your diagnostic skills in a patient with progressive weakness and fever (James Milburn *et al*) as well as in a clinicopathological report of a patient presenting with cognitive decline (Shona Scott *et al*, *see page 466*).

Diagnosis is essential but is only the beginning. Randomised controlled trials (RCTs) are major undertakings for drug treatments but are fantastically difficult and unusual for surgical treatments. Thymectomy has seemingly helped patients with myasthenia for many years and finally we have results from the RCT originally set up by John Newsom-Davis. Jon Sussman summarises the trial's findings and sets them in the context of the ABN myasthenia gravis guidelines.

Many of you kindly completed our Reader Survey earlier this year, and we summarise the findings on *page 428*. The resulting word cloud fortuitously had a Christmassy feel.

Competing interests None.

Provenance and peer review

Commissioned; internally peer reviewed.



Figure 1 Word cloud from reader survey of results of question ‘how has Practical Neurology impacted on your work?’