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Highlights from this issue

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Common diseases are common. Rare diseases are rare. Or rather, rare diseases are individually rare, but taken together they are quite common, at least in neurology. Neurologists are often accused of having spanophilia (the love of the rare),¹ as if it is inevitably a failing, but given the very large number of rare diseases in neurology it is almost a requirement for neurologists. And probably a good thing, as long as it does not get in the way of common sense. In this edition we let loose our inner spanophile and highlight why spanophilia is important for the practical neurologist—as well as for their patients.

Rare disease can be difficult to diagnose yet may respond to specific treatments, which clearly makes these diagnoses very important. Primary angiitis of the central nervous system illustrates this well, discussed by Shamik Battacharyya and Aaron Berkowitz (see page 195); in another example, Thomas Webb and colleagues diagnosed *Listeria meningoencephalitis* in a roundabout way (see page 220). Porphyria is one of those rare diseases that you consider in a wide range of situations—but what do you do when you have a patient with it? Simona Balestrini and colleagues explore their approach to difficult management decisions in one of their patients (see page 217).

There is a standing joke between the editors that anything novel in *Practical Neurology* must be a misprint. Certainly we do not aim to publish new reports—with novelty comes uncertainty and we prefer content with some resilience. However, we are keen to make our readers aware of newly described conditions, particularly treatable disorders. We were therefore

delighted to receive two reports of a recently described condition—IgG4-related disease—by Claire Rice and Monica Marta and their colleagues (see pages 235, 240). This flurry of submissions does make us wonder if it will turn out to be rather less rare than it currently appears.

But what of rare untreatable diseases, the sort of thing that historically has given neurologists a bit of a reputation? Karlien Mul and colleagues provide helpful hints on how to diagnose facioscapulohumeral muscular dystrophy (see page 201) and Gillian Ingram and colleagues describe a patient with an unusual motor neuropathy whose singing problems led to a genetic diagnosis (see page 247). Mary Reilly provides an articulate and persuasive editorial that explores the benefits that an 'exact genetic diagnosis' can bring for the patient and for the doctors looking after them, even without available specific treatments (see page 174).

Looking in the mouth is a part of the standard neurological (and general) examination but is diagnostically rewarding only relatively infrequently. Sashank Prasad and colleagues report a pair of cases where this examination was very useful (see page 231).

We have two reviews from paediatric neurology colleagues. When children with cerebral palsy grow up, they consult adult neurologists. Their diagnosis has already been made and management plans are usually in place. Neil Wimalaundera and Valerie Stevenson provide a helpful review to highlight when to reconsider the diagnosis and which management options to think about. Many interventions in paediatrics are based not upon trials in

children but on inferences from trial results in adults. The opposite is true for ketogenic diets for epilepsy. These diets can be very helpful in selected patients, even though their use becomes more difficult as children get older. Natasha Schoeler and Helen Cross provide a practical approach to using these diets in adults (see page 208); this is our editors' choice so is free to distribute to patients, adult dieticians and other colleagues.

Disorders of visual perception are unusual and relatively rare and the phenomenology is of interest to the general public (as in 'The man who mistook his wife for a hat')² as well as to neurologists. Sarah Cooper and Mike O'Sullivan review the phenomenology and associated anatomy and pathophysiology (see page 176) to keep you several steps ahead of the reading public. Oliver Sacks was a champion weightlifter whilst practising neurology and would doubtless have appreciated Marion Simpson's description of her own weightlifter's headache (see page 215), a presentation that presumably is still a rarity within our profession.

Common things are common so we must think about unusual presentations of common diseases before focusing on rare diseases. But playing the percentages only goes so far: for the patient with that rare disease, its prevalence is 100%.

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REFERENCES

- 1 Asher R. *Talking sense about medicine*. London: Psychology News Press, 2015.
- 2 Sacks O. *The man who mistook his wife for a hat*. London: Duckworth, 1985.