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FOCUS ON FOCAL EPILEPSY

Extraordinary claims need extraordinary evidence; which is why it is always reassuring when a major breakthrough is reported simultaneously by two groups. The discovery of *DEPDC5* as a cause of 12% of familial epilepsy with variable foci represents a significant advance in the understanding of the focal epilepsies. This is a prime example of exome sequencing unravelling the aetiology in families too small for conventional linkage analysis and discovering a biologically plausible gene—but a gene where we know relatively little regarding its function.

Nature Genetics 2013 (Epub ahead of print)

Nat Genet 2013 (Epub ahead of print)

AGE RELATED ATHEROSCLEROSIS

The modern plagues of obesity and smoking are drivers of the atherosclerosis that is all too prevalent, right? Well perhaps not. An imaging study of 137 mummified corpses across four distinct populations (ancient Egypt, ancient Peru, the Ancestral Puebloans of south-west America, and the Unangan of the Aleutian Islands) identified that atherosclerosis was common 4000 years ago. It was identified in 34–60% of the groups studied. Whether those who were mummified represented all peoples at that time is unclear; we can only speculate as to whether they were living a ‘contemporary lifestyle’ of indolence and excess.

Lancet 2013 (Epub ahead of print)

I SENSE IT IN MY WATERS

How do you identify neurodegeneration before it becomes irreversible? CSF biomarkers and genetic studies attempt to do this, but a recent project combined the power of both. A genome-wide association study of 1269 people with Alzheimer’s looked to provide an answer. Three loci were associated with tau/ptau CSF levels: near *GEMC1* and *OSTN* (3q28); *GLIS3* (9p24.2); and the *TREM* gene cluster (6p21.1). The first of these loci strongly associated with clinical and pathological markers of dementia in an independent data series.

Neuron 2013 (Epub ahead of print)

PROFESSOR PLUMB, WITH THE LEADING PIPING, IN THE LIBRARY...

When considering ‘man-in-the-barrel’ syndrome—consider whether instead of an oak cask, it may instead be a lead drum. Pickrell and colleagues report in the *Lancet* a rapidly progressive brachial diplegia associated with microcytic anaemia and basophilic stippling (figure 1). Blood lead levels were over nine times the upper

limit of normal which prompted a ‘Dr House’ style investigation of the person’s property and the discovery of lead drinking pipes in their remote rural dwelling. Chelation therapy produced a dramatic and satisfactory clinical resolution.

Pickrell *et al. Lancet* 2013;381:1156

IN FLIGHT UPTHURST

One of the PN Editors recently visited the land of the long white cloud—no doubt because of the cutting edge research that is bubbling under in this fine nation. Pommergaard and colleagues proffer the definitive opinion on a research question that has baffled generations of scientists—what do to about flatulence on airplanes. In short they suggest ‘just let it go’—but go on to advise the use of activated charcoal (in either trousers or the seat cushions) to neutralise any associated odour. Chocks-away!

N Z Med J 2013;126:68–74.

► Additional references are published online only. To view please visit the journal online (<http://dx.doi.org/10.1136/practneurol-2013-000586>).



Figure 1 Incontrovertible bilateral wrist drop.