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THERE'S NO PLACE LIKE EXOME

Rare genetic disorders pose a conundrum to the clinician—it can be tricky to know where to start when commissioning gene tests. An international consortium performed exome sequencing on 84 individuals with progressive myoclonus epilepsy; a third of cases showed mutations likely to be causative in known genes. More interestingly, a further 11 unrelated people had the same de novo mutation (p.Arg320His) in *KCNC1*—which had not previously been linked to epilepsy. De novo mutations are interesting here as the symptoms were relatively late-onset and because the progressive myoclonus epilepsies were conceptualised as mostly recessive. The ‘diagnostic exome’ when it comes to a lab near you may help to illuminate these rare but important diagnoses.

Nat Genet 2014. doi:10.1038/ng.3144

WISDOM COMES WITH SIZE NOT WRINKLES

Why some people are more likely develop a cognitive impairment than others is a complex and often a somewhat sensitive question. An increasing body of evidence from neuroimaging suggests that general cognitive ability may relate to both the degree of an individual's cortical folding and total cortical area. These two metrics are clearly related but it is not clear which is more important. A recent detailed study investigated the relationship between cognitive ability and various metrics derived from MRI in 512 male twins aged 51–60. Genetic factors explained nearly 85% of cortical folding, and both the degree of cortical folding and cortical surface area significantly correlated with cognitive ability.

However, the relationship between cognitive ability and cortical surface area remained significant having controlled for gyral folding, but not vice versa. This suggests that increased gyral folding—and therefore shorter anatomical connectivity between brain regions—does not explain cognitive ability. What does underpin this relationship is still an unanswered question.

NeuroImage 2014. <http://dx.doi.org/10.1016/j.neuroimage.2014.11.040>

PUBLISH AND BE DAMNED

Medical academics with low self-esteem publishing in an unglamorous field may console themselves that at least two people will read their paper: the Editor and themselves. A Fo Ben has unfortunately found two examples where even that predicted readership was a clear overestimate.

To err is human, and there are many days that A Fo Ben feels very human indeed. However, the authors of ‘Variation in Melanism and Female Preference in Proximate but Ecologically Distinct Environments’ published in *Ethology* must have wanted the ground to open up and swallow them whole when an internal comment between authors ended up published in the full article. That the line was ‘(should we cite the crappy Gabor paper here?)’ makes it all the more alarming that peer review, proof reading and inhouse editorial mandarins allowed this through.

There are many online-only, open-access journals; this is a legitimate field of publishing. However, there are predatory low-quality journals that spam scientists offering to publish their work for a fee. One such example is the *International Journal of Advanced Computer Technology*

who accepted and published a manuscript from David Mazieres and Eddie Kohler which had a pithy seven-word title. Controversially, the entire paper was these seven words repeated without variation from Abstract to Conclusion—‘Get me off your F**ing Mailing List’. Readers who do not baulk at profanity should be guided to the figures used to illustrate their 10-page magnum opus.

<http://www.theguardian.com/australia-news/2014/nov/25/journal-accepts-paper-requesting-removal-from-mailing-list> (Guardian newspaper 25 November 2014).

Ethology 120:1090–1100.

FROM SCAN-ROOM TO COURTROOM

Perhaps the inevitable, if unintended, consequence of the proliferation of papers over recent years suggesting that brain imaging can tell us almost everything about how the brain works is the increasing use of functional imaging as evidence in legal cases. Increasingly, neuroscientists are being asked to acquire these data and interpret results in the courtroom. In a recent ‘how-to’, four experts in the field discuss the pitfalls of attempting to explain these complex techniques in a legal setting, and what to think about, should you be called on to interpret these findings. This is an interesting, and increasingly important, read which frames what most of us probably know about the limitations of these techniques in a rather different light.

Nat Rev Neurosci 2013;14:730–6.



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