Carphology by A Fo Ben

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NONSENSE AND SENSIBILITY

Educationalists ask us to identify our learning style: are you an abstract conceptualist or an active experimenter for example? Well if you learn best by having disorders mapped on to classic works of literature, with a tongue simultaneously in cheek, you are in luck. The superbly frivolous 'Pride and Protein' from William Stern reinforces the presentation and inheritance of the ornithine transcarbamylase deficiency. In doing so, he may have explained the curious paucity of males in the Bennet family at the centre of Austin's *Pride and Prejudice*, and more importantly that half of the females are sensible, while half are silly. The clue, writes Stern, is that silliness is more likely at family gatherings—pointing to a defect in protein metabolism.

J Inherit Metab Dis 2016;39:321–4.

SWAN LINK

What does it take to identify 14 news genes associated with neurodevelopmental disorders? The exomes of 4293 families and a further scouring of 3287 published cases. The scale of the Deciphering Developmental Disorders is a testament to the British and Irish researchers who collaborated on this project. Cases were recruited in busy clinical genetics groups and were children with undiagnosed neurodevelopmental disorders. Predominantly, these were 'SWANs' (syndromes without a name). They confirmed the effect of increasing paternal age on the likelihood of a child having an important new gene mutation. Forty-two per cent of cases had a pathogenic new mutation in a coding gene; half of these disrupt gene function and the rest alter protein function. The automatisation of phenotyping permitted the creation of composite photographs for the facial gestalt for many of these new syndromes.

Nature 2017;542:433–8.

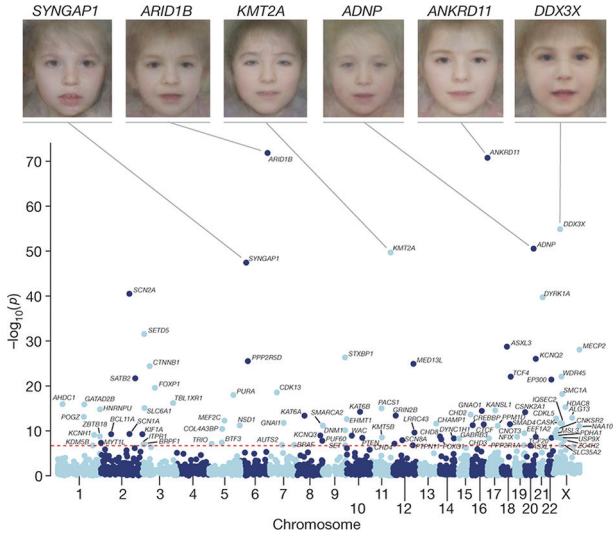

Figure 1 Manhattan plot of combined p values across all tested genes. The red dashed line indicates the threshold for genome-wide significance ($p<7\times10^{-7}$). Composite faces were generated from clinical photos of individuals with new mutations in the same gene. Taken from Deciphering Developmental Disorders Study. Prevalence and architecture of de novo mutations in developmental disorders. *Nature* 2017;542:433–8.

BREAKING THE FOURTH WALL

A Fo Ben is the pseudonym used by more than one writer of Carphology since we took over from Rajendra 8 years ago (February 2009). The name has a tenuous origin, deriving from a line from the *Mabinogion* (a collection of 12th to 13th century myths and folk stories from North Wales). In one tale, Bronwen and her men find themselves in a bit of a pickle, so Bendigeidfran stretches out over the river to make good their escape, exclaiming *'A fo ben, bid bont'*. This can be translated as 'Let he who would be a leader, be himself a bridge'. It is our aim to be a bridge to the knowledge squirrelled away—predominantly outside of the major neurology journals.

We had no expectation that Google Scholar, the academic data-scraping engine attached to the search behemoth, would struggle so much with knowing how to reference these articles. Unsurprisingly, they are often credited to 'AF Ben', but the engine often misattributes authorship to the title of the first story. Hence, such scrambled authorship gems as 'TY Memory' (2009), 'DNA Junk' (2012) and 'K Nailgun' (2010). The piece that gave Google Scholar the biggest headache began 'Men are from Pars reticular and women are from the venous sinus' (yes, I know) and was apparently co-written by the duo of Menaref Pars and Waref Reticula.

Pract Neurol 2010;10:62; *Pract Neurol* 2014;14:70; *Pract Neurol* 2012;12:400; *Pract Neurol* 2009;9:310.

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