DIAGNOSING NORMALITY...

Our medical school graduation year book opened with the apocryphal quote that ‘There’s no such thing as a well patient, merely one who hasn’t been investigated enough’. Partially tongue in cheek, it nonetheless resonated with me even then. I had already noted as a student that with a bit of time and observation many ‘cases’ could be ‘de-diagnosed’ or (semantically better) ‘re-diagnosed’ as variants of the normal (and indeed Normal) phenotype. Martin Ward Platt’s editorial (see page 915) encapsulates this eloquently. This desire to infer pathology, appears to be taking a stronger hold, the result perhaps of well meaning, but overzealous guidelines, a loss of the concept of ‘caseness’ and (and we can debate this) a societal malaise in which the default is now ‘abnormality’ where it once was the opposite.

This month’s issue includes several papers which address this theme through different lenses. All are brave in that they tackle an issue many would prefer left alone.

CORONARY ARTERY DISEASE PART 1: THE TRANSIENT VARIANTS

The study by Coon and colleagues (see page 937) illustrates the phenomenon perfectly. Their hypothesis is that Kawasaki’s disease and coronary artery abnormality (CAA) outcome might fit the overdiagnosis syndrome. This could have been catalysed by changes in recommendations to more intensive echocardiographic vigilance despite most CAs being benign and spontaneously resolving. Their interrogation of a US database (the Paediatric Health Information System) with primary diagnoses of Kawasaki’s disease between 2000 and 2014, showed that while there was no change in severe coronary artery abnormality (CAA) outcomes (the ones that really matter), the rate of diagnosis of any CAA more than doubled over the era. Though exact scan details are not available on this system, it is hard to come to any other conclusion that the more you look (and of course higher resolution scanners whet the appetite for the search), the more of unimportance one finds.

DE-HOSPITALISING INVESTIGATION

It is well known that children with Down syndrome are at high risk of obstructive sleep apnoea which, in turn can influence general health, cognitive performance and demeanour. The gold standard diagnosis, a multichannel plethysmographic recording involves hospital admission and is time consuming, expensive and often unpalatable to families. Though it is ‘recommended’, like much guidance, evidence is in short supply. Hill and colleague (see page 962) validated the use of a simple one night home saturation study using readily-derived variables. A combined measure incorporating parameters easily measured this way (desaturation, delta 12 s indices and 3% oxyhaemoglobin) performed extremely well with a sensitivity of 92% and specificity of 63%, suggesting that half of all inpatient studies could be avoided by a simple, acceptable home alternative.

HYPOADRENALISM: NOT NECESSARILY LONG TERM

Transient biochemical abnormalities in neonates are well recognised as is the fact that most self-resolve. A number of babies with hyponatremia and hypoglycaemia are genuinely hypoadrenal, but, do we really know what a minor deviation in a Synacthen provocation means given the small numbers studied and inherently wide variances in testing in this age group? A neat study by Tan and colleagues (see page 984) demonstrates how easy it is to mistake normality for permanent endocrinopathy. In their group of 68 babies with this biochemical phenotype, about 40% had an ‘abnormal’ ACTH stimulation. Of these, however, 70%, had no cause identified, and in this group, 90% simply resolved over a few months. The message in this case is not ‘don’t investigate’ but to ‘keep an open mind’ and be prepared to de-diagnose the baby with a pathological label.

GLOBAL HEALTH: CASH TRANSFER PROGRAMMES

In existence for decades, cash transfer programmes in low and middle income countries (LMICs) and high income countries (HICs) have much more in common, their neo-liberal philosophy (responsibility with the individual) underpinning each. In HICs they may be called ‘welfare’ or ‘social assistance’ programmes and can be conditional (on employment) as in North America or unconditional, the prevalent form in Scandinavia. In LMICs, conditional programmes tend to depend on engagement of families with primary education and health surveillance. Siddiqi’s ambitious systematic review (see page 920) sought to identify areas of similarity and difference between schemes though effect sizes in these studies are notoriously hard to measure and even harder to summate. Though the number of papers small, there was a suggestion that conditional programmes in HICs did not alter behavioural patterns, while in LMICs (like the Mexican Opportunidad programme) they tended to change both engagement and outcomes positively. Like all Global Health papers, this piece is free.

CORONARY ARTERY DISEASE PART 2: THE PERSISTENTLY ABNORMAL

In the era of pumps and continuous glucose monitoring, it would be easy to become complacent about insulin dependent (‘type 1’ in old parlance) diabetes. Though recent changes in management may, in time, prove to have attenuated the risk, Cheung and colleague’s analysis of the risk of death from ischaemic heart disease in children and young people registered on the Yorkshire database between 1978 and 2014 deaths is something of a wakeup call (see page 981). Through careful scrutiny and validation of the death certificates, they estimated a standardised mortality ratio of 13.8 in this group. This figure is impossible to attribute to normal variation, but would have been underestimated without the extra interrogation.

We seem to have come full circle and perhaps need to be as good at ruling out pathology in as out, or more specifically knowing when to look. I’m sure there’s an aphorism that fits the bill there too.