



Highlights from this issue

Nick Brown , *Editor-in-Chief*

As the first anniversary of the first adult COVID-19 cases approaches, we reflect on what we've learned since December 2019 and could be excused a frisson of excitement about what the soon-to-be-released vaccines might offer. More on this story soon of course, but here are some thought provoking non-corona papers.

NEXT GENERATION SEQUENCING

It seems only a few minutes since the CGH array was being heralded as the great diagnostic saviour after the limitations of the 'traditional' karyotype and deletion detection methods were recognised. Next generation sequencing, based on refinements on technology introduced by Sanger in the 1970s has now effectively supplanted all that came before to the extent that it is finding use (or being touted for use) in rapid, 'bedside' diagnostics (metabolic to dysmorphology) as well as the better known outpatient work up approach. Diana Baralle's editorial on the science behind NGS (including whole exome and whole genome sequencing) adds to two studies from Singapore, Neha Bhatia and Heming Wei in which additional diagnostic yield in children in whom traditional methods have been negative. Both studies found positives in the 35% to 40% range, higher in certain phenotypes (neuromuscular and skeletal dysplasia) universal additional information for counselling and results which often changed treatment. *See pages 1, 31 and 38*

Global child health

Snakebite: management

Jay Halbert and Jacqueline Le Geyt continue their brilliant series on snakebite, this instalment reviewing management. Never has *primum non nocere* been more germane, much harm being (unwittingly) caused by traditional 'cures'. Primary treatment is generic to all species and includes: non-weight bearing and simple analgesia; immobilisation of the bitten part of the body so it lies below the level of the heart;

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referral to a medical facility with attention to the airway, oxygenation and prevention of aspiration and gaining intravenous access in an unaffected limb. Harmful practices such as incision, suction devices, snake stones, cryotherapy and tourniquets are now known to be high risk. Tourniquets can increase local tissue destruction and cause gangrene. Pressure immobilisation bandages are useful in bites by elapids (neurotoxic snakes that do not cause local swelling) to reduce lymphatic flow but can cause harm in viperid bites and are therefore not recommended by WHO in most snake bites. If the snake type has been identified (not always possible—photos can help) then anti-venom specific to the family of the biting snake can be added. This treatment is specific to the type of bite, the coagulopathy of the *Viperidae* or the neurotoxicity of the *Elapidae* families. *See page 14*

Epinephrine auto-injectors: gentle or jabbing?

There are two schools of thought as to the optimum way of administering emergency epinephrine with an auto-injector for anaphylaxis: the gentler place and press method and (possibly faster) method of swing and jab. Confusingly, different devices recommend one or the other, while some (eg, EpiPen) recommend both depending on geographical region. Louise Pike and David Tuthill assess whether there are other gains from the use of one method over the other, using the length of (paintball drawn) laceration from needle-free practice pen tests as a marker for trauma and pain in a group of Welsh primary school children. The place and press technique 'incurred' far less of a mark, suggesting less real-life risk of a laceration and a more pleasant experience (if that's an appropriate term given the use to treat anaphylaxis). For sheer pragmatism and ingenuity, this is my editor's choice for the month. *See page 54*

Non alcoholic fatty liver disease

In a compelling review of non alcoholic fatty liver disease (NAFLD), precursor to NASH, steatosis, Meera Shaunak explores the pathophysiology and potential interventions. The folkloric perception of the obesity equation has now been debunked: it is one part of the equation, but dietary composition (UFAs, disaccharides)

and chronic hypoxia and ethnicity all contribute. Intervention is extremely difficult, the usual arsenal of metabolic-modifying drugs (metformin, losartan, anti-oxidants), so far in the ' tantalisingly promising' rather than clearcut delivering phase. *See page 3*

Thyroid anatomical phenotypes

Though thyroid imaging after a diagnosis of congenital hypothyroidism (CH) is deemed 'desirable', the use of scintigraphy (a much more sensitive tool for detection of variants in position) has yet to become embedded in the routine work up, partly as many are yet to be convinced that it changes management. Chris Worth's analysis of a 10 year (2007–2017) study of neonatal CH/ TSH screen positive babies might change this view. In their series, scintigraphy was routine and more babies with gland in situ (GIS) and gland ectopia and fewer a/dysplastic glands than expected found. Those with GIS had lower median TSH and higher LT4 than their counterparts and a high chance of the hypothyroidism being transient (off treatment by 3 years of age) and it feels as if scintigraphy has untapped potential as a prognostic tool. *See page 77*

Cycle of deprivation and abuse

Though the use of electronic records is ubiquitous, there is still much untapped potential. Identifying households at high risk of intimate partner violence and child maltreatment from 'precursor' warning presentations is one example of their promise. Shabeer Syed and colleagues' systematic review of test validation studies eruditely pools the positive predictive values for a range of warning diagnoses (fractures, abstinence syndrome in children for example) and later ascertainment/corroborator. With the (unsurprising) rider of publication bias, markers had between 50% and 90% PPV, the only low outlier being fetal alcohol syndrome, a notoriously difficult diagnosis even when directly reported. Somehow (through data set linkage) these flags need to be translated to warning systems: if not, we will have missed a major opportunity. *See page 44*

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