



# Highlights from this issue

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Genomics is becoming integral to diagnosis, treatment and prognosis of a disease. Various articles in this month's issue highlight the rapid embedding of genomics in clinical practice. The systematic application of genomic technologies is making precision medicine a true possibility. Precision medicine is an innovative approach for disease treatment and prevention that considers individual variability in genes, environment and lifestyles for each person. Christopher Hine *et al* (see page 379) provides a simple overview of how small molecule precision medicine (CFTR modulators) have been developed and applied. The article describes how this innovation is revolutionising cystic fibrosis care and providing potential genotype-specific treatment options for about 90% of cystic fibrosis patients. The article takes us on a journey from discoveries in fundamental sciences to its translation into clinical practice and improved patient outcomes. This article is the Editor's choice of the month, as it has a real world impact and provides an insight into the paradigm shift which clinicians need to swiftly understand and embrace.

Even if genetic results do not suggest a treatment, they are still meaningful to families. Irnthu Premadeva *et al* (see page 338) explores key factors that should be considered during withdrawal of neonatal intensive care. The authors explore the practicalities and psychology involved. They highlight the important role of clinical genetics in difficult decision-making and in providing answers to grieving families. Genetic information accrued can also inform management of future pregnancies. Prompt access to diagnostic

tests like whole genome and exome sequencing has made identification of inherited monogenic disorder possible in these difficult situations. However, although genetic variations are now more readily identifiable, these tests may still not be able to provide answers. Lisa J Bryson and Shelagh Joss (see page 383) provides an insight on how to use genetic testing after sudden infant death syndrome. The authors emphasise discussing testing with a clinical geneticist when the family history and case pathology suggest a possibility of genetic causation. Strict classification of pathogenicity and the need for further research into functional analysis of relevant genetic variants is also highlighted.

The above articles all focus on a personalised approach to patient care. The diametrically opposed model of standardising care delivery also has merit, as in the excellent example given in article by Nicholas Embleton. He provides a simple, practical ABCDE approach to nutritional assessment in preterm infants (see page 314). This is a worthwhile read, as the author explores key concepts and principles which guide nutrition in preterm infants. The epigenetic effects of dietary components on cognition and metabolic outcome are well-known. A consistent, cohesive approach to nutritional assessment can positively influence these outcomes. The guidance provided is easily adaptable to be applicable at the bedside. It also allows input from all healthcare professionals and families looking after vulnerable infants.

This month's edition revisits Adrian Plunkett's work on positive approach of learning from excellence. The article 'Embracing

excellence in healthcare: the role of positive feedback' (see page 351) highlights the importance of widening learning potential across the whole landscape of work, from successes to failures. The idea that we can learn from everyday success is a feature of Safety-II,<sup>1</sup> where safety is considered a situation in which as many things as possible go right, rather than a situation in which as few things as possible go wrong. The author discusses practical steps to embrace excellence and positive feedback, including using an appreciative inquiry approach to consolidate learning and generate improvement ideas.

Positive feedback for excellence, can offset the negativity associated with blame which tends to hinder our ability to learn from failures. This nurturing of positive culture within team and individual can lead to improvement in self efficacy and resilience. A cultural change in workplace to complementary approach is even more important concept today's environment where for a number of complex reasons, morale in the medical profession is low and stress is high.

One of the key aspects for a paper to appear in a print is peer-review process. Our reviewers provide their expertise and time generously. They are the unsung heroes of the journal. I would especially like to express my gratitude for their contributions in this issue.

Hope you will enjoy reading this month's papers and continue contributing enthusiastically.

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## REFERENCE

- Hollnagel E, Wears RL, Braithwaite J. From Safety-I to Safety-II: a White Paper 2013.