The Genome Revisited

Editorial

The field of Medical Genetics has been growing by leaps and bounds. We have come a really long way since the initial description of heritable traits by Gregor Mendel in the 1860s. The elucidation of the human genome sequence, which would have seemed like science fiction until even half a century back, has become a reality and now we are actually looking at ways of modifying it through genome editing techniques. The explosion of information about the human genome has been paralleled by the tremendous advances in genetic testing technologies. The genetic diagnostic armamentarium has been rapidly expanding, beginning with the development of the karyotyping technique for chromosomal analysis in the 1950s to the evolution of the Next generation sequencing technology in the 21st century. Each technique when introduced, opens up a world of diagnostic possibilities, only to be overshadowed by a more advanced technology with an even more fascinating scope and a higher diagnostic yield.

The chromosomal microarray (CMA) technology, which began to be used extensively in clinical practice from the late 2000s onwards, was one such development. CMA made genome-wide screening of copy number variations possible at a much higher resolution than conventional karyotyping, so much so that it became the recommended first-line genetic test for the evaluation of unexplained intellectual disability and uncharacterised multiple malformation syndromes. It has retained its pre-eminent position as the molecular cytogenetic test-of-choice for almost a decade now. But it seems as though it is going to be gradually replaced by the newest entrant in the arena – Whole genome sequencing (WGS). The GenExpress in this issue summarizes three recently published studies by Halgren et al., Zhou et al. and Van Opstal et al., which have demonstrated the superiority of WGS over CMA in the elucidation of molecular cytogenetic aberrations in the prenatal as well as postnatal scenario. WGS can detect CNVs at a far higher resolution than CMA and can help to exactly map chromosomal breakpoints down to the last nucleotide.

As technology continues to evolve, we are going to keep revisiting the genome, delving further into its intricacies and unearthing more and more of its secrets. Therefore, it is essential for all of us, working in the field of Medical genetics, doctors and scientists alike, to stay abreast of these rapid developments. Genetic Clinics endeavours to play a small but significant role as a travel guide for this journey through the human genome.

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1st October, 2018