

Preface

Medicine is at the crossroads as we march into a new genomic era, brought about by major technological advances. The disruptive technology of next generation sequencing holds exciting promise for disease gene discovery, improved understanding of the biological bases of many genetic disorders, which for some will translate to targeted therapies, and for many families will lead to much improved genetic counselling.

Genomic studies in children and adults have the potential to integrate genomic medicine to primary healthcare practice and bridge the gap between the basic research and clinical application. Early detection of cancer using specific biomarkers from genomic studies has been shown to increase the detection rate, improve treatments with less treatment-associated morbidity, and enhance the quality of life of affected individuals.

Now that the treatment of infectious and nutritional disorders is well established, pediatric onset genetic disorders and cancer in children constitute a substantial load in pediatric clinic nowadays.

In this special edition of the “genomic studies of paediatrics” of the *Translational Pediatrics* we have focused on the topics of genetic disorders and cancers, with 11 articles selected to review the recent advances in our understanding of a range of genetic diseases

Phenylketonuria (PKU) has an incidence of 1 in 10,000 in the general population. Dr. Gladys Ho and Professor John Christodoulou have reviewed the current knowledge about PKU, and how current genomic research is being translated into novel therapies for the disorder.

Dr. Kaustuv Bhattacharya, Tiffany Wotton and Associate Professor Veronica Wiley (the current President of the International Society of Neonatal Screening) have reviewed the past evolution of newborn screening, discussed the current state of the art, and predict what the future might hold for newborn screening.

Stem cell research is one of the fastest developing areas in medical research. Dr. Aiqun Wei and colleagues discussed the recent advances in the capacity to differentiate mesenchymal stem cells into a diverse range of specialised cell types, and suggest that such research might potentially provide hope for disc regeneration. In this review a number of controversies, challenges and future therapeutic concepts were also considered.

According to the WHO, infertility and sterility caused by genetic and non-genetic factors are predicted to be the third most serious disease worldwide in the 21st century after cancer and cardiovascular diseases. Assisted Reproductive Technology (ART) is an approach to treating such diseases and for enhancing the reproductive options for carriers of genetic disorders. Dr. Jie Qiao and Associate Professor Hui-Liang Feng reviewed the related government policies in China in the ART field, and consider them both in the Chinese and global context.

Uniparental disomy and aneuploidy caused by Robertsonian translocations can cause neuromotor developmental delay in children. Dr. Moh-Ying Yip has proposed a testing strategy for parental carriers of Robertsonian translocations and individuals with congenital anomalies.

Dr. Yue Huang and colleagues reviewed the state of the art genetic testing for four major areas of hereditary neurological disorders in children, including recommendations for investigation and interpretation of results.

Preterm birth is an important health issue in neonates, with both genetic and non-genetic factors contributing to its pathogenesis. Dr. Qin Zhu and colleagues have explored the role of inflammatory cytokine gene single nucleotide polymorphisms in the geographic and racial variability of preterm birth.

Cell free DNA technologies, both in research and clinical applications, have developed very quickly over the last few years. Dr. Shuye Wang and colleagues have reviewed these advances, and their applications in both cancer and non-cancer domains.

The complex analysis of genomic rearrangements as part of the diagnostic work-up of leukemia can be difficult using routine haematological studies and the conventional karyotype. Dr. Bo Guo and colleagues have described a technology that enables the detection of the complex genomic rearrangements in leukemia, which is an important requirement for determination of effective treatments.

Dr. Xin Wang has reviewed the measurement of the DNA index of the genomic rearrangements, using flow cytometry, to identify biclonality at diagnosis, and to monitor minimal residual disease in pediatric leukemia to assist clinicians in tailoring of therapy.

Dr. Federrica Saletta and colleagues have reviewed progress in pediatric cancer treatment over the last decade, and have provided an overview of seven novel therapeutic agents which have become particularly popular, and have reported on the progress of clinical trials by using these agents.

We would like to thank all authors for their excellent contributions to this special issue of *Translational Pediatrics*, having drawn on their experience and expertise. We also would like to thank reviewers, Nancy Q. Zhong and to their members of the *Translational Pediatrics* editorial team for their effective and diligent work. We hope this special edition will be of interest to those who have an interest in these research fields.



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