Molecular Syndromology

Themed Issue: Craniosynostosis

Editor: Erwin Pauws (London)

Preface

Understanding craniosynostosis: clinical practise to basic science

While the first surgical correction of craniosynostosis was reported by Lannelongue in the late 19th century, it took more than a hundred years before the first causative gene for this condition was identified. Since then, great progress has been made towards the elucidation of the genetic basis of craniosynostosis for many patients, providing an accurate molecular diagnosis for themselves and their families. Over the last few decades, the discovery of more than 50 genes causing craniosynostosis has helped to clarify the pathophysiology of the disease, which in turn, has led to improved clinical care. At the same time, detailed studies into the biological function of genes and proteins associated with craniosynostosis have significantly advanced our understanding of the disease pathogenesis. These advances have not only been able to inform and enhance current treatment regimens for craniosynostosis, but have also identified molecular and cellular mechanisms that underpin the normal development and homeostasis of the craniofacial skeleton. As a result, the prospects of developing novel treatments for craniosynostosis are now starting to become a reality. This is particularly important because surgical correction of craniosynostosis and the consequential secondary defects puts a heavy burden of care on patients and of course, their families.

This special issue of Molecular Syndromology contains both review articles and original research articles that describe the current state regarding clinical practice, genetic diagnosis and associated underlying pathogenesis of craniosynostosis. Together they highlight the latest and most significant developments in the field. To my knowledge this is the first themed journal issue on craniosynostosis and I hope that this collection of papers will be able to interest experts in the field as well as attract new researchers, bringing novel ideas and impetus to this enthralling aspect of craniofacial malformation.

I am grateful to all contributing authors, referees and to the editorial support from Michael and Karen Schmid.

Cover figure legend

Left: Sagittal section of the coronal suture showing the neural crest-mesoderm boundary in detail. The parietal bone (p) is of mesodermal origin (red) while the frontal bone (f) is of neural crest origin (green). **Right**: Functional gene network containing genes associated with craniosynostosis.

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