Foreword

Genetic Advances in Craniofacial Malformations

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It is with great pleasure that we have organized this special issue of *Journal of Pediatric Genetics* focusing on “Genetic Advances in Craniofacial Malformations.” The care of patients with craniofacial anomalies dictates expertise from various disciplines, including genetics, plastic surgery, otolaryngology, dentistry, orthodontics, speech and language pathology, among others. In addition, this patient population is diverse, and patients may have isolated anomalies or present with a constellation of features suggestive of specific genetic syndromes.

Most craniofacial anomalies are malformations or morphological anomalies resulting from an intrinsically abnormal developmental process. Malformations may have genetic, environmental, or multifactorial etiologies. The knowledge of the genetic etiologies of craniofacial malformations is rapidly evolving in a large part due to the recent advances in next generation sequencing technologies. In addition, genomics research identifying susceptibility loci for a variety of craniofacial malformations is ongoing as well as research related to the study of gene–environmental interactions.

The current issue contains a series of five articles reviewing recent advances in several fields of craniofacial biology. The articles cover a broad range of anatomic findings, includ-

ing the eye, the ear, the jaw, the teeth, and others. In the article by Plaisancie et al, the authors review the genetic etiologies of anophthalmia and microphthalmia. In the article by Gendron et al, the vascular, environmental, and genetic causes of microtia are reviewed. Ahmed et al, reviews jaw malocclusions and discusses genetic syndromes presenting with different malocclusions. In the article by Ye et al, non-syndromic and syndromic origins of tooth agenesis are reviewed. Finally, Yoon et al examines genetic screening in patients presenting with craniofacial malformations.

A recurrent theme in this issue, which highlights the complex nature of craniofacial malformations, is the necessity of a multidisciplinary approach. In our center, similar to others, patients are examined by multiple disciplines at single patient appointments and discussed during team conferences. This approach aids in the identification of a uniform, specific diagnosis, and a concerted treatment plan. In the future continued interaction between specialties will become even more important as newer diagnostic modalities appear and additional craniofacial genetic syndromes are identified.

It is with great pleasure that we present this special edition.

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