Neural tube defects (NTDs) are among the most common serious birth defects. They include a spectrum of malformations that can be lethal (e.g., anencephaly, craniorachischis), associated with significant disability, morbidity and mortality (e.g., spina bifida, encephalocele), or result in more modest impact on function and quality of life (e.g., lipomeningocele). While some NTDs can be prevented by maternal periconceptional use of folic acid supplements or consumption of folic acid fortified foods, only a small proportion of NTDs worldwide are prevented by such strategies (Youngblood et al. 2013). Furthermore, despite over 30 years of serious inquiry, the mechanism(s) by which folic acid protects against NTDs remains unknown. Similarly, the mechanisms linking NTDs with the few known risk factors (e.g., maternal diabetes, maternal use of specific medications) are largely unknown, and despite strong evidence of a genetic component, there are no confirmed genetic risk factors for NTDs in humans.

Initiated in 1999, the International Neural Tube Defects Conference series brings together researchers from a wide variety of disciplines who have the common goal of understanding and ultimately of preventing NTDs. The Eighth International Neural Tube Defects Conference, held October 7 to 10, 2013, was organized by Richard Finnell, Laura Mitchell, Gary Shaw and John Wallingford, and supported by generous contributions from Pamlab and Zeiss. Ninety attendees from 13 countries (United States, Australia, Canada, Chile, China, Egypt, France, Italy, Malaysia, Nicaragua, Qatar, Spain, and the United Kingdom) convened at the AT&T Executive Conference Center in Austin, Texas. In addition to the superb conference facilities, the venue provided easy access to the vibrant academic and social environs of the University of Texas as well as some great Texas barbeque!

Since their inception in 1999, the NTD Conferences have provided a forum for the exchange of information across the fields of clinical medicine, public health and the basic sciences. Following in this tradition, the Eighth International NTD Conference included 44 oral and 21 poster presentations in the broad categories of clinical research, embryology and development, genetics and epigenetics, environmental risk factors, folate and other nutrients and emerging technologies. In addition, Dr. E. Marcotte, Professor, Department of Molecular Biosciences at the University of Texas at Austin, provided a keynote address on "Systems Biological Approaches to Neural Tube Defects", in which he described experimental and computation strategies for discovering disease-relevant systems of proteins.

The papers included in this issue of Birth Defects Research (Part A) provide a glimpse of the interdisciplinary nature of the NTD conferences, as well as the spectrum of research being conducted by the vigorous NTD research community. Appropriately, the first paper in this issue (Kancherla et al., 2014), which documents the increased mortality experienced by individuals with the spina bifida, reminds us of the importance of research directed toward improved NTD prevention. Several papers that explore the mechanisms underlying observed associations with NTDs are also included in this issue, including a comprehensive review of the evidence linking NTDs to mitochondrial one-carbon metabolism (Momb and Appling, 2014). In addition, Sanders et al. (2014) describe an embryonic stem cell model for neural tube development in a diabetic environment and Fathe et al. (2014) describe their use of ELISA assays and cell culture modeling to explore a novel mechanism by which valproic acid may cause spina bifida.

The importance of animal models in studies of neural tube development is highlighted by a paper describing a new spontaneous mouse mutant, tuft, which is characterized by a lipomatous cephalocele (Fong et al., 2014), and studies in the null knock out mouse model of Gpr161 (Hwang and Mukhopadhyay, in press). In addition, Herion et al. (2014) review mouse genetic models to identify the molecular pathways that, if perturbed, may result in mesodermal insufficiencies and structural malformations including NTDs. Finally, Hansler et al. (2014) describe a metabolic profiling platform that can be used to identify metabolic changes associated with NTDs in early stage mouse embryos. Moving to humans, De Marco et al. (2014) provide an overview of their studies assessing the association between NTDs and genes in the planar cell polarity pathway and Krupp et al. (2014) consider the potential impact of next-generation-sequencing on our understanding of the genetic contribution to NTDs in humans.

Marcy Speer (1959–2007) was one of the initial founders of the International NTD Conference series. At each
meeting, Marcy is remembered and honored through the Marcy Speer Memorial Awards, which are given to the pre-doctoral and postdoctoral level trainees who are judged to have provided the most meritorious platform presentations. The awardees for the 2013 meeting were Juliette Petersen (doctoral), Howard Hughes Medical Institute, University of Colorado Denver, for her presentation, “Towards an Understanding of the Role of Folic Acid in Neural Tube Closure”, based on work conducted in the laboratory of Lee A. Niswander, and Dr. Ran Blekhman (postdoctoral), now an assistant professor at the University of Minnesota, for his presentation, “Using Deep Whole-Genome Sequencing to Understand the Role of Non-Coding Genetic Variation in Complex Developmental Disease”, based on work conducted in the laboratory of M. Elizabeth Ross, Weill Cornell Medical College.

The International NTD Conferences provide an interdisciplinary forum for sharing current research on the causes of NTDs. These meetings have helped to foster existing collaborations, served as a catalyst for new collaborations, and provided trainees with the opportunity to identify postdoctoral and faculty level positions within groups with shared research interests. Planning for the Ninth International NTD Conference, once again to be held in Austin, Texas, is currently underway. Details will be posted to: http://ntdconference.com/ when they become available.

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References


