

NICHD Sixth Structural Birth Defects Meeting

May 13-14, 2008

The Conference Center at the Maritime Institute, Linthicum Heights, MD

Summary

The Sixth Structural Birth Defects Meeting sponsored by the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) and the National Institutes of Health (NIH) Office of Rare Disease was held at the Conference Center at the Maritime Institute in Linthicum Heights, MD on May 13-14, 2008. It was attended by principal investigators funded through the NICHD's Birth Defects Initiative, Program Staff from NICHD, the National Heart Lung and Blood Institute, and the Centers for Disease Control and Prevention (CDC) as well as Scientific Review Officers from NICHD and the NIH Center for Scientific Review (CSR).

The purpose of the meeting was to bring together currently funded researchers whose work focuses on the developmental mechanisms or the genetic susceptibility and variability of human structural birth defects. This working group is unique in that it includes both basic scientists and clinicians, and the projects they oversee emphasize the translational aspects of birth defects research. The meeting provided a forum for the investigators to discuss the plans and progress of their research, communicate and interact, exchange ideas and information, share resources, and foster synergistic collaborations that will enhance the research goals of the NICHD Birth Defects Initiative.

The scientific presentations provided updates on research on the molecular mechanisms underlying Cornelia de Lange Syndrome and the cohesinopathies; neural tube defects (spina bifida); congenital contractures, isolated clubfoot, scoliosis, and skeletal dysplasias (achondroplasia, brachyomia, spondyloepimetaphyseal dysplasia, bent bone dysplasias, osteogenesis imperfecta); pediatric hormone deficiency diseases; defects arising from ciliopathies; and, congenital diaphragmatic hernia. Dr. Margaret Honein of the National Center on Birth Defects at the CDC presented an update on the National Birth Defects Prevention Study and highlighted opportunities for collaboration with the nine sites funded as part of this study which has completed over 30,000 maternal interviews and processed nearly 14,000 infant DNA samples. Dr. Toni Scarpa, Director of the NIH CSR, discussed current efforts to revamp the review process at NIH particularly the review of clinical and translational research applications, an issue of relevance to the Birth Defects Working Group. Finally, during the Town Hall meeting the group discussed the next meeting (to be held in fiscal year 2010) and the pros and cons of piggybacking onto another meeting of a relevant scientific society. Because the members of the Working Group find the meeting so stimulating and useful, the possibility of applying for a Gordon Conference focused on structural birth defects was also discussed as a potential mechanism to expand the group.