Guest Editorial

Neurobiology of Disease in Children: Neurofibromatosis 1

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Scientific advances of the past decade have improved our understanding of the pathogenesis of many disorders characterized by neurologic dysfunction. In several of these disorders, new diagnostic tools and therapies have been developed, tested, and adopted. However, the pace of discovery in the basic and clinical neurosciences has made it challenging for child neurologists to determine what molecular diagnostic strategies, sophisticated neuroimaging tools, or innovative therapies should be adopted to manage not only common ailments but also the estimated 1500 rare diseases that cause neurologic dysfunction.

In 2001, the National Institutes of Health funded a 5-year conference series titled “Neurobiology of Disease in Children.” The overall goal is to bring together clinicians, scientists, caregivers, and National Institutes of Health program officers to determine how research findings can be translated to enhance clinical understanding and affect clinical practice.

Neurofibromatosis 1 was selected as the topic for the first symposium. The conference format included formal presentations, question-and-answer sessions, panel discussions, and open discussions to directly address conference objectives. The symposia directors were Drs Bruce Korf and Peggy Wallace, who are highly respected leaders in the field of neurofibromatosis. The panel included speakers and moderators, all experts in their own fields, selected by the directors to ensure authoritative presentations on key topics and productive discussions on research directions. The question-and-answer sessions after each set of talks encouraged brainstorming between the panelists and audience of child neurologists about the best directions of future research and how clinicians can contribute.

The first presentation, “Current Approaches to Diagnosis and Molecular Testing,” described current approaches to the diagnosis of neurofibromatosis 1 and progress toward the development of molecular genetic testing. The second session, “Clinical Manifestations in Context of Pathogenesis,” addressed the pathophysiology of neurofibromatosis 1 through a review of various laboratory approaches. These presentations provided the audience with an understanding of current concepts in protein function, tumor suppressor mechanisms, and tumor biology as they relate to neurofibromatosis 1 features. The topics in the third session, “Cognitive Aspects of Neurofibromatosis 1,” related to the central nervous system effects of neurofibromatosis 1 mutations and neurofibromin deficiency, such as learning disabilities, and provided a description of the animal models that allow understanding of subsequent cognitive and cellular effects. The fourth session, “Current Therapies and Clinical Trials,” discussed approaches for therapy, current or proposed clinical trials, and questions germane to planning such trials (eg, natural history, measurement of outcomes). The last session was an open discussion moderated by Drs Robert Finkelstein and Giovanna Spinella, which provided an opportunity for clinical and basic scientists to define future research directions in neurofibromatosis 1.

I wish to express my sincere appreciation to the National Institute of Neurologic Disorders and Stroke (NINDS), the Child Neurology Society, Neurofibromatosis, Inc., and the National Neurofibromatosis Foundation for cosponsoring the conference, which was attended by more than 150 child neurologists.

One of the specific aims of the “Neurobiology of Disease in Children” conferences is to disseminate the proceedings of the symposia to ensure that clinicians and basic scientists are informed about scientific advances, current research initiatives, and future directions. This issue of the Journal of Child Neurology features a series of articles prepared by conference participants and the transcript of the conference’s question-and-answer sessions, panel discussions, and open discussions.

Leukodystrophies have been selected as the topic for the second “Neurobiology of Disease in Children” conference, which will be held on October 9, 2002, in Washington, DC. In keeping with the objectives of the original proposal, the program directors, Drs Hugo Moser and Sakku Naidu, have prepared a superb agenda that includes a panel of the most highly respected leaders in the field of leukodystrophies. We look forward to seeing you in Washington!