

**5th Meeting of the Down Syndrome Consortium:
A Public-Private Partnership**
July 11, 2013

**6100 Executive Boulevard
5th Floor Conference Room
Rockville, Maryland**

Updates

Dr. Maggie Nygren, American Association on Intellectual and Developmental Disabilities (AAIDD), is actively involved in amicus briefs in death penalty cases involving individuals with disabilities.

Dr. George Jesien, Association of University Centers for Disabilities (AUCD), had an advisory committee call with the National Center for Prenatal and Postnatal Down Syndrome Resources in collaboration with Lettercase, a nonprofit that has developed website for parents (<http://downsyndromediagnosis.org/>) and is working on the leadership institute at University of Delaware. Their staff just met with Karen Summar (representing Rep. Cathy McMorris Rodgers).

Dr. Michael Harpold, Down Syndrome Research and Treatment Foundation (DSRTF), reported that DSRTF is holding many events around the country and working to raise awareness for research. During its recent grants process, DSRTF made \$1.2 million in grants for coming year. Its drug research is showing some improvement in cognition (repurposing). There will soon be an announcement about new research.

Michelle Livingston, Global Down Syndrome Foundation (GDSF), reported that GDSF is hosting a webinar with the Texas Department of Aging and Disability Services. Tom Blumenthal and Huntington Potter spoke about new tools being used in this research and the science behind the connection with Alzheimer's disease. GDSF will host a Research Roundtable Day at next week's meeting on July 18.

Mitchell Levitz, self-advocate at the University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development, reported that the center held focus groups on advocating as young adults. He and George Jesien are on the same board.

Deanna Tharpe, Down Syndrome Affiliates in Action (DSAIA), reported that DSAIA is revamping its website, with a full resources page, with the goal of launching by October 1. A Request for Proposals link is ready for those interested in presenting at DSAIA's Washington, D.C., conference in February 2014.

Dr. Bob Riddle, National Institute of Neurological Disorders and Stroke (NINDS), reported that everything that NINDS has been doing has been part of the trans-National Institutes of Health (NIH) working group.

Dr. Charlene Schramm, National Heart, Lung, and Blood Institute (NHLBI), had no updates.

Dr. Lisa Gilotty, National Institute of Mental Health (NIMH), reported that NIMH has reissued a program announcement for research. NIMH and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) are collaborating on an activity in the fall.

April 2013 DS/Alzheimer's Meeting

Dr. Melissa Parisi, NICHD, provided an overview of meeting. It built on Kathleen Gardiner's meeting that occurred just prior, which had been funded by NIH and several other organizations.

Dr. Michael Harpold, DSRTF, asked whether any of the Down Syndrome Consortium (DSC) members could provide input. He mentioned that several Requests for Applications are currently out that might apply to both DS and Alzheimer's, and he would like for DSC members to suggest some initiatives.

Dr. Yvonne Maddox, NICHD, suggested that since this is an important statement, the DSC might want to put a notice in the NIH guide to alert the DS community to these opportunities. Melissa Parisi, NICHD, reported that NICHD will put together a summary of the workshop and could post that on the DSC website.

DS Registry Update

Yvonne Maddox, NICHD, reported lots of positive feedback. Operations and governance boards have met. NICHD could post the membership of these two boards, but it should ask people whether they want their names on the website.

Melissa Parisi, NICHD, provided an update and overview on its status. There will be an Institutional Review Board meeting at the end of July to review the consent form, the formal protocol, and the intake survey. NICHD is creating and reviewing additional survey modules on specific health issues. Its goal is to launch in mid-to-late August. NICHD requested external scientific review from four reviewers who gave excellent feedback. Regarding clearance from the Office of Management and Budget, NICHD is optimistic that it can get a waiver. It is holding monthly calls with the operations board and quarterly calls with the governance board. It now has an initial health questionnaire. NICHD would like to have a booth at the National Down Syndrome Congress meeting to invite families to try out the registry and give feedback. To date, 25 beta testers have tested the site.

Yvonne Maddox, NICHD, floated the name "DS-Connect™: The NIH Down Syndrome Consortium Registry."

Vanessa Rangel Miller, PatientCrossroads, noted that the goal of the PatientCrossroads home page is to invite a user in. PatientCrossroads might solicit some videos and make it interactive. The different informational sections are under development. It is not accessible to the public currently.

Ms. Miller walked the DSC members through the registration process: After you "join the registry," you create an account in a secure manner. PatientCrossroads used streamlined consent language (Section 508 compliant) with consent options and a security code to prevent false registrations. The dashboard allows access to tools in the system, and multiple surveys can be listed. You can update account information, upload scanned records, and do more. The user mostly has preset response lists (e.g., types of DS); some are required questions, because they are key elements. Users can start survey, pause, and resume later. Only the user has access to the password for his or her account. PatientCrossroads will never share identifying or contact information. The registry will share de-identified summary data.

Discussion of New Members of the DSC

Yvonne Maddox, NICHD, reported that NICHD has received recommendations for possible new additions to the DSC: **Mark Bradford** of the Jérôme Lejeune Foundation, which supports research on therapies and treatment; and **Brandy Helleard** of the International Mosaic Down Syndrome Association (IMDSA). NICHD does not want too large a group but needs important input. NICHD received an email from Colleen Jackson-Cook, a cytogeneticist who chairs an IMDSA advisory group, asking whether she can dovetail her data.

The general consensus from the DSC was that these were two good organizations to include, but it was suggested that each organization have an opportunity to comment. These new members would have the same type of involvement as current members.

Discussion of Potential Funding Models

Yvonne Maddox, NICHD, reported looking into co-funding grant applications that might miss the NIH payline. NICHD will also talk with the trans-NIH DS Working Group, hoping to provide opportunities to fund more DS research, as: (1) a gift fund designated for DS; or (2) having program officials contact the Primary Investigators who miss funding to tell them that some organizations might be able to fund them in fiscal year 2013.

DSC members discussed Dr. Maddox's comments and also suggested (1) providing links to organizations that provide research grants and (2) pooling resources to help pay for science. Lisa Kaeser and Mary Lou Oster-Granite will bring this discussion up with the trans-NIH DS Working Group and make a presentation at the DSC's next meeting. DSC members can help with prioritization.

Attendees

Lisa Gilotty, NIMH

Michael Harpold, DSRTF

George Jesien, AUCD

Lisa Kaeser, NICHD

Mitchell Levitz, self-advocate, University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development

Michelle Livingston, GDSF

Yvonne Maddox, NICHD

Maggie Nygren, AAIDD

Mary Lou Oster-Granite, NICHD

Melissa Parisi, NICHD

Vanessa Rangel Miller, PatientCrossroads (guest)

Bob Riddle, NINDS

Charlene Schramm, NHLBI

Deanna Tharpe, DSAIA