7th Meeting of the Down Syndrome Consortium:
A Public-Private Partnership
November 21, 2013

6100 Executive Boulevard
5th Floor Conference Room
Rockville, Maryland

Updates

Dr. Yvonne Maddox, *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), noted Dr. George Jesien's retirement from Association of University Centers on Disabilities (AUCD), lauding him for his many accomplishments on behalf of people with disabilities.

Dr. Michael Harpold, Down Syndrome Research and Treatment Foundation (DSRTF), announced the award of 2013–2014 research grants, totaling $1.2 million and focusing on cognition, intellectual and developmental disabilities, and Alzheimer's disease. A research meeting is being held in Cambridge March 27-29, 2014, on Alzheimer's and Down syndrome (DS). There is potential for international collaboration.

David Tolleson, National Down Syndrome Congress (NDSC), gave an update on the database and infrastructure that NDSC is developing. NDSC is holding its annual convention in Indianapolis in July 2014 and in Phoenix in June 2015. It is partnering with the Global Down Syndrome Foundation (GDSF) on several fronts. NDSC is very interested in efforts toward diversity and would like to discuss further; it wants to reach out to families who are not registering.

Mitchell Levitz, self-advocate, University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development, attended the AUCD conference, including the pre- and postnatal review board meeting.

Michelle Livingston, GDSF, reported that GDSF has new Alzheimer's grants, totaling $1.2 million, in conjunction with the Alzheimer's Association and the Chicago Adult Clinic. Dennis McGuire recently retired but will consult with GDSF and the adult clinic in Denver. They want to grow it into a version of the Sie Center for Down Syndrome (almost 800 patients). GDSF is working with the National Down Syndrome Society, NDSC, and the Jérôme Lejeune Foundation (JLF) on a congressional briefing, which will provide updates on research, education policy, and the Achieving a Better Life Experience Act. Representatives Pete Sessions, Eleanor Holmes Norton, and Cathy McMorris Rodgers hope to attend. It will be open to Senate staff.

Deanna Tharpe, Down Syndrome Affiliates in Action (DSAIA), reported that DSAIA is getting ready for the conference in Washington, D.C., on February 28, 2014. She would like an update from NIH. The new DSAIA website is up. She inquired about the timing of the next possible Down Syndrome Consortium (DSC) meeting.

Brandy Hellard, International Mosaic Down Syndrome Association (IMDSA), reported that IMDSA is rolling out an e-conference in March 2014. It will cost $21 for 21 webinars over the course of 3 days, designed for families, on topics ranging from Individual Educations Plans to research (http://www.321econf.net). IMDSA's retreat weekend will be in Williamsburg, Virginia, on October 3–5, 2014, focusing on research.

Mark Bradford, JLF, reported that JLF will participate in the congressional briefing. Its international scientific conference will be in Paris in October 2014. JLF will announce new grants soon.

Dr. Bob Schoen, Research Down Syndrome (RDS), reported that RDS announced 2013–2014 grants 2 months ago. RDS gave a special award to Dr. Maddox and Dr. Roger Reeves at its D.C. event.

Dr. Laurie Ryan, National Institute on Aging (NIA), reported that NIA held a DS and Alzheimer's meeting in June 2013. The meeting saw possibilities for partnerships, similar to the Alzheimer's Disease Neuroimaging Initiative, and NIA will be holding more discussions. Melissa Parisi added that the executive summary will be available very soon and will become part of the DS research plan.

Dr. Charlene Schramm, National Heart, Lung, and Blood Institute (NHLBI), reported that NHLBI continues to support grants, including to Children's Hospital in Boston, that look at how progenitor cells do and do not form blood cells. NHLBI is also researching functional modeling for upper airway disorders and how it relates to obstructive sleep apnea.

Dr. Lisa Gilotty, National Institute of Mental Health (NIMH), spoke on the importance of the current Funding Opportunity Announcement on trying to understand mental illness in individuals with intellectual and developmental disabilities.

DS-Connect™ Update

Melissa Parisi, NICHD; Sujata Bardhan, NICHD; and Vanessa Rangel Miller, PatientCrossroads
Presentation attached

- The new flyer is posted on the website as a PDF; the group requested that DSC members download and use it. The group has already distributed at meetings, and local groups are using it. Dr. Maddox suggested that the flyer be put on members' websites as well.
- There is a new "Need Help?" tab on the homepage.
- DS-Connect™ has 690 new registrants in November, for a total of 1,136 in the United States.
- The main sources of visitors to the registry site are NIH, Facebook, NDSC, and DSRTF. Ms. Miller stated that emails that include the registry link also are a major source, along with results from search engines. NIH followed up with emails to NDSC convention attendees.
- The group showed the map of current registrants. The Midwest is doing great, and a few registrants are international.
- The demographics of the registrants are about evenly split between males and females but skew younger (884 under age 30).
- The group used congenital heart defects as an example of how they can pull de-aggregated data out.
- The group reviewed other features under development: leukemia, the Professional Portal, the ability to print responses, customized messages, and a newsletter.
- The group requested feedback from members on outreach plans, video testimonials, how-to videos, and further dissemination.

Discussion

Many Consortium members expressed sentiments that good work had been done on the registry. Other ways to spread the message about the registry were suggested: (1) Do a webinar at the 321 e-conference and other webinars in Spanish; (2) put information in all children's hospitals; (3) reach out to the American Congress of Obstetricians and Gynecologists to pass information on to expecting mothers; (4) target other places where DS groups gather, especially adults with DS; and (5) post testimonials on the registry site. DSC members also agreed that communications need to be uniform and to emphasize the security of the site and the information.

NICHD solicited members to join subcommittees to help with activities such as outreach. Several members identified themselves as willing to help: Ms. Tharpe, Mr. Bradford, Ms. Livingston, and Mr. Tolleson.

Research Plan Update

Lisa Kaeser and Dr. Mary Lou Oster-Granite, NICHD, provided an update on the DS Research Plan. The draft Research Plan should be posted after Thanksgiving. They asked the DSC to share the draft widely with their members and will send a notice when it has been posted.

Discussion and Next Steps

NICHD noted that it was considering issuing a Request for Information to stakeholders to find out how they might use the data. A member suggested adding a professional portal and frequently asked questions (FAQs) to the registry to help engage the research community as much as possible before the meeting in the United Kingdom in March. Other discussion centered on submitting information or a commentary to leading journals as a way to raise awareness for the registry.

Attendees

Sujata Bardhan, NICHD
Tom Blumenthal, Linda Crnic Institute for Down Syndrome
Mark Bradford, JLF
Kyle Brown, PatientCrossroads (guest)
F.L. Dammann, NICHD
Lisa Gilotty, NIMH
Michael Harpold, DSRTF
Brandy Hellard, IMDSA
Colleen Jackson-Cook, IMDSA
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, American Association on Intellectual and Developmental Disabilities
Mary Lou Oster-Granite, NICHD
Melissa Parisi, NICHD
Vanessa Rangel Miller, PatientCrossroads (guest)
Bob Riddle, National Institute of Neurological Disorders and Stroke
Laurie Ryan, NIA
Robert Schoen, RDS
Charlene Schramm, National Heart, Lung, and Blood Institute
Deanna Tharpe, DSAIA
David Tolleson, NDSC