9th Meeting of the Down Syndrome Consortium:
A Public-Private Partnership
July 12, 2014

6100 Executive Boulevard
5th Floor Conference Room
Rockville, Maryland

Welcome and Introductions
Melissa Parisi, Eunice Kennedy Shriver *National Institute of Child Health and Human Development (NICHD)*

Update on DS-Connect™
Presentation attached

Dr. Melissa Parisi, NICHD, provided the update:
- The National Institute on Minority Health and Health Disparities (NIHMD) has joined as a new member. Dr. Yvonne Maddox, former chair of the Down Syndrome Consortium (DSC), is the new Acting Director. LuMind and the Special Olympics Committee have new logos; these have been updated on the DS-Connect™ website.
- Dr. Parisi described the structure of the registry, including the operations and governance boards and the registry coordinating team. A new registry review committee is being formed to oversee researchers' requests to recruit registry participants.
- Photos showed the registry team and the operations and governance boards. These will be posted on the website to build trust among potential participants.
- A map showed where current registrants are located around the United States.
- There are still more males than females registered. The majority of registrants are younger than age 20.
- Dr. Parisi demonstrated an example of how to use the "Explore the data" graphics.
- Dr. Parisi noted that NICHD has focused on families who want to contribute but also want something back, such as a list of health care providers, growth measurements, connections with resources about Down syndrome (DS), and assurance that information is secure. She mentioned that new Centers for Disease Control and Prevention-sponsored updated growth charts for DS will shortly become available and would be embedded on the website.
- The listing of health care providers will include their specialties as reported, but with the disclaimer that the NICHD cannot rate these professionals.
- After registrants fill out the registry survey questions, they can click on a PDF version and print out a summary statement that can be used when meeting with health care providers.
- Regarding the need to reassure potential registry participants that their information will be secure, DS-Connect™ has the Federal Information Security Management Act moderate level of security, which means that no social security information or bank information are gathered, and the registry is regularly audited.
- Updated frequently asked questions are being posted now by the Information Technology team. Lisa Kaeser will send them to DSC members as soon as they are up.
- The Professional Portal is in process. The health care professionals who access the DS-Connect™ site will have no direct access to registry participants; there will be three levels of data access with three levels of review and approval. A new data registry review committee will review levels 2 and 3. NICHD will also include a requirement that professionals submit letters of support for their projects.

- It is easy to open an account on the Professional Portal. It requires a point of contact and asks why the health care provider would like access to the registry.
- Dr. Parisi demonstrated how to search for particular conditions and ages and how many individuals match those search criteria. If too few people are registered in a particular location, the request will bounce back to help protect their privacy.
- Future developments include new questionnaire modules on female health, cause of death, and leukemia. NICHD is also working on a glossary, a Spanish-language version, expanded resources, and health care reminders.
- The goal is to link to biospecimen repositories using Global Unique Identifiers (GUIDs).

Discussion

DSC members discussed the recent Heartbleed computer virus and the emails that were sent regarding the virus. There were no security breaches of the DS-Connect™ website but users are able to change their passwords if they had any concerns. Additionally, users will be required and notified to change their passwords every 6 months. It was also noted that the health care provider list is private and cannot be used to link back to a registrant.

Outreach Efforts

Dr. Sujata Bardhan, NICHD, provided an update, asking DSC members for their input:

- DS-Connect™ has 1,930 registrants with a goal of 10,000.
- Each DSC member present was given a flash drive with the electronic outreach toolkit, which includes: badges; info cards; flyers; a health care provider form; Microsoft PowerPoint slides; and full-size images that can be used in your own presentations, on Twitter, and on Facebook. A ZIP file would also be sent to all members with this information.
- There are different info cards for use with multiple audiences.
- All of these resources are free.
- The DS-Connect™ team is currently developing video testimonials that will speak directly to parents and individuals.
- Dr. Bardhan asked DSC members to sponsor how-to webinars, use the e-toolkit, publicize DS-Connect™, and get the message out.
- Deanna Tharpe suggested having everyone promote the registry at their walks, including the takeaway cards and flyers (a PDF on the registry website).

Discussion

The rate of registration was discussed. Other disease registries usually see a 10 percent to 15 percent rate in the first year. The DS-Connect™ team has reached out in a variety of ways (webinars, poster sessions, local seminars, the podcast, and a radio blog), but some meetings focus on families with younger children. At the Down Syndrome Medical Interest Group (DSMIG) meeting, many health care professionals provided their information for the Professional Portal. Moving forward, the DSC should think of other strategies to reach additional groups. The flyers currently used at meetings explain the registration process and are formatted for someone with DS. The idea of an app was also discussed.

Other DSC Business

Lisa Kaeser, NICHD, provided an update on the Research Plan. It will be published after it is made Section 508 compliant.

An issue was raised about whether and, if so, how to add new members to the DSC, including federal agencies as ex-officio members.

The group discussed term limits and decided that each organization should determine who would best represent them and for how long.

Updates

Deanna Tharpe, Down Syndrome Affiliates in Action (DSAIA), reported that in April, DSAIA launched a new accreditation program to grow its membership. It is currently planning the next annual conference in Las Vegas on March 6–8, 2014. The speaker Request for Proposal is posted online.

David Tolleson, National Down Syndrome Congress (NDSC), reported that NDSC has held many side meetings at the conference and appreciates the National Institutes of Health (NIH) and DSMIG. Next year, the conference will be held in Phoenix in June. NDSC has hired a new person for outreach to African American families to address cultural barriers. Mr. Tolleson recommended an adult toolkit for DS-Connect™ to help facilitate discussions within families and to help siblings as parents age.

Dr. Maggie Nygren, American Association on Intellectual and Developmental Disabilities (AAIDD), reported that AAIDD will distribute the DS-Connect™ flyer to every attendee at its upcoming conference.

Michelle Sie Whitten, Global Down Syndrome Foundation (GDSF), reported that GDSF is surveying medical professionals to help with updates to its prenatal testing pamphlet and is announcing grants to local organizations. GDSF is planning a November conference and conducting a national survey with adults. The Sie Center for Down Syndrome has a quarterly research publication and holds educational seminars for parents and professionals.

Dr. Bob Schoen, Research Down Syndrome (RDS), reported that RDS recently sent letters to six new grantees to announce their awards.

Sara Weir, National Down Syndrome Society (NDSS), reported that NDSS is holding a congressional event next Wednesday with Representatives Pete Sessions and Cathy McMorris Rodgers. NDSS has established a Discovery Fund to support clinical infrastructure. The Achieving a Better Life Experience Act is getting close to passage on Capitol Hill.

Dr. George Capone, DSMIG, reported that DSMIG held its 5th continuing medical education conference yesterday, focusing on registry, biobanks, and databases. It also held its first strategic planning session. With membership increasing, DSMIG wants to find ways to support community-based providers and may try to create a toolkit. It has an increasing presence from Latin America.

Mark Bradford, Jérôme Lejeune Foundation (JLF), reported that the Governor of Pennsylvania is signing the Prenatal Education Act on Friday. JLF has four new grant recipients. New projects are coming, including an application in development to stay connected. JLF expects results next week for the Barcelona study and is working on medical guidelines for adults and a new clinic in Paris.

Dr. Marilyn Bull, American Academy of Pediatrics (AAP), reported that the DS guidelines are being automatically reviewed in 2014 by the AAP Committee on Genetics. AAP is trying to get the message out to 64,000 pediatricians. It has had ongoing communications with the Special Olympics Committee, and its new medical guidelines will recommend that everyone get a medical exam for their spinal cords. AAP should do another news article when the Professional Portal is launched.

Mitchell Levitz, self-advocate, University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development, noted an online training module for teens and young adults and will share the link when it is ready.

Dr. Colleen Jackson Cook, International Mosaic Down Syndrome Association (IMDSA), reported that NIH will be presenting at the IMDSA October research conference. On Saturday of that weekend, people participate in research projects (e.g., studies of depressive symptoms, cognition, sleep, and mitochondria). IMDSA is putting data together to focus on mosaic DS but welcomes others with non-mosaic DS. It will hold its 321 spring conference in 2015 and would like to expand to self-learning modules.

Dr. Tom Blumenthal, Linda Crnic Institute for Down Syndrome (LCI), reported that the institute is helping researchers add a DS module to their current grants. LCI has 23 labs at the University of Colorado working on DS now, as well as five Alzheimer's and DS grants. LCI is setting up a new society on Trisomy 21 research with Roger Reeves and a postdoctoral fellowship aimed at DS research. The institute wants to add collaborations between the university and other researchers, and Dr. Blumenthal noted that these efforts will help train the next generation of DS researchers.

Attendees

Sujata Bardhan, NICHD
Tom Blumenthal, LCI
Mark Bradford, JLF
Marilyn Bull, AAP
George Capone, DSMIG
Melissa Parisi, NICHD
Michael Harpold, LuMind, formerly the Down Syndrome Research and Treatment Foundation
Colleen Jackson Cook, IMDSA
Lisa Kaeser, NICHD
Maggie Nygren, AAIDD
Vanessa Rangel Miller, PatientCrossroads (guest)
Bob Schoen, RDS
Deanna Tharpe, DSAIA
David Tolleson, NDSC
Sara Weir, NDSS
Michelle Sie Whitten, GDSF
Mitchell Levitz, self-advocate, University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development (by phone)