4th Meeting of the Down Syndrome Consortium:
A Public-Private Partnership
March 14, 2013

Note: This meeting of the Down Syndrome Consortium (DSC) was hosted by the Congressional Down Syndrome Caucus on Capitol Hill.

Welcome and Introductions
Karen Summar

Updates
Rep. Cathy McMorris Rodgers recognized that Dr. Yvonne Maddox, Deputy Director of the *Eunice Kennedy Shriver* National Institute on Child Health and Human Development (NICHD), spoke at the very first caucus briefing. Unlocking the mysteries of the 21st chromosome will affect people with Down syndrome (DS) and many others. Moving research in that direction has a multiplier effect.

Dr. Yvonne Maddox, NICHD, updated the caucus on NICHD and research at the National Institutes of Health's Institutes and Centers. Dr. Maddox explained the makeup and intent of the DSC and its research and support efforts, including research opportunities, cost sharing, the launching of the DS patient registry, the work on a database, and the biobank. The registry could not have been accomplished without the DSC.

Dr. Charlene Schramm, National Heart, Lung, and Blood Institute (NHLBI), reported that NHLBI has supported grants for projects that include growing cells in lab, including those from people with DS, allowing for research that cannot be done on humans, and upper airway modeling on computers to study precise anatomy of breathing difficulties in DS genes and how they affect the severity of atrial ventricular defects.

Dr. Neil Buckholtz, National Institute on Aging (NIA), reported that NIA has been researching what happens to DS individuals as they age, especially its relationship with Alzheimer's disease. NIA is funding clinical trials in older individuals and antibody studies in Alzheimer's and DS. DS is part of the National Alzheimer's Project Act, to be released in May 2012. NIA is finding ways to track the recommendations. On chromosome 21, amyloid protein, leading to plaques, become more plentiful in DS patients as they get older. A recent advance is the discovery of radioactive chemicals that bind to the amyloid in the brain. Now we can follow people over time to see what happens; before, we could only measure after people had died.

Dr. George Jesien, Association of University Centers on Disabilities (AUCD), spoke of current programs and projects, including 15 Intellectual and Developmental Disability Research Centers (e.g., the University of Wisconsin language development program). Vanderbilt University is looking at strengths-based approaches and visual-spatial relationships and will be doing a webinar on the Towson research in which 10 individuals with DS did much better on a test than people without DS. Vocational rehabilitation has denied training in the past for people with DS; that may change soon. The Bright Futures program is producing more balanced information about living with individuals with DS. DS and fragile X syndrome both have effects mitigated by certain drugs. A California foundation has issued three small grants for adult health care for individuals with disabilities.

Dr. Melissa Parisi, NICHD, gave an update on the DS registry and introduced the registry subcommittee: Jon Colman, National Down Syndrome Society (NDSS), and herself. The registry awarded a contract in September 2012 to PatientCrossroads, which has experience with the Global Rare Disease Registry.

Dr. Michael Harpold, Down Syndrome Research and Treatment Fund (DSRTF), reported that DSRTF is focusing on DS cognition research on therapies, studying neuropathology of dementia and cognitive decline. DSRTF has eight potential therapeutics. It has a new paradigm-changing strategy, allowing it to integrate various projects together. DSRTF is involving pharmaceutical companies, has several studies approaching clinical trials, and has supported work (e.g., Edgin, Nadel) to develop an Arizona cognitive test battery validated for individuals with DS. This provides a tool to measure whether a therapeutic works. DSRTF has two trials on new Roche drugs, pentylenetetroazol (NICHD and the National Institute of Neurological Disorders and Stroke [NINDS]) and an investigational new drug, to address Alzheimer's.

Dr. George Capone, Down Syndrome Medical Interest Group (DSMIG), represented a group that includes 100 to 150 people who provide direct care to individuals with DS. It provides continuing medical education credits for training. DSMIG is developing guidelines for overweight, obesity, and sleep apnea. It is important to know whether standard therapies work, as comorbidities have varying impacts on families. DSMIG feels that it must understand what is successful for our families, including the economic impact of treating or not treating.

David Tolleson, National Down Syndrome Congress (NDSC), reported that NDSC, which provides information and advocacy for people with DS and their families, held its 41st convention this year. It raised issues around the Achieving a Better Life Experience (ABLE) Act of 2013 and the Trisomy 21 Research Resource Act of 2011.

Mitchell Levitz, self-advocate at the University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development, is on the review committee for prenatal and postnatal DS, a member of AUCD, and a co-author of Count Us In. He updated the DSC on his project, which created an iPad application.

Deanna Tharpe, Down Syndrome Affiliates in Action (DSAIA), represented a trade association that provides resources and training for local DS organizations, families, and clinicians. DSAIA collects and disseminates information.

Jon Colman, NDSS, reported that NDSS is expanding its Washington, D.C., operation with Sara Weir. While NDSS does not directly fund research, now it is taking steps toward doing so. Mr. Colman came for the Buddy Walk. NDSS wants to pass the ABLE Act.

Michelle Livingston, Global Down Syndrome Foundation (GDSF), reported that GDSF straddles both worlds, improving lives through education and research. It supports the Linda Crnic Institute for Down syndrome. In the past 6 months, GDSF has granted $1.3 million to 14 researchers through the University of Colorado. Huntington Potter is building the first Alzheimer's center at the university. GDSF announced in July a joint grant initiative with the Alzheimer's Association. From the clinical care perspective, GDSF is growing and performing more outreach. It sees 600 patients with DS at its Colorado site, providing care and translational research. GDSF is excited about registry. It will hold a Research Medical Day on July 18 in Denver, providing a free half-day of clinical care and half-day of research.

Dr. Cara Long, NINDS, reported that the NINDS portfolio focuses on cognitive impairments. NINDS is looking at specific genes on chromosome 21 to identify targets for potential interventions.

Lisa Kaeser, NICHD, announced the website launch and explained features, including links to all DSC members, the registry landing page, and a Trisomy 21 Awareness Month spotlight.

Dr. Melissa Parisi, NICHD, reported that on April 13–15, NICHD held a workshop on DS cognition, with broad sponsorship by DSC members.

Attendees

Neil Buckholtz, NIA
George Capone, DSMIG
Jon Colman, NDSS
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
Cara Long, NINDS
Yvonne Maddox, NICHD
Rep. Cathy McMorris Rodgers, U.S. House of Representatives
Melissa Parisi, NICHD
Charlene Schramm, NHLBI
Karen Summar, staff, Rep. Cathy McMorris Rodgers
Deanna Tharpe, DSAIA
David Tolleson, NDSC