

**6th Meeting of the Down Syndrome Consortium:
A Public-Private Partnership**
August 23, 2013

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

Welcome and Introductions

Yvonne Maddox, Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

The launch of the Down Syndrome (DS) registry is August 28.

Dr. Maddox welcomed new members to the Down Syndrome Consortium (DSC):

- **Dr. Colleen Jackson-Cook**, International Mosaic Down Syndrome Association (IMDSA), is delighted to participate. She noted that families want to move the research forward.
- Brandy Hellard, IMDSA, noted that this furthers the IMDSA mission regarding mosaic DS and mosaic translocation.

Update on DS-Connect™: The Down Syndrome Registry

Melissa Parisi, NICHD, and Vanessa Rangel Miller, PatientCrossroads

The tab for professionals is still under development but should be ready within about a month. Anyone with an account can explore the data. The news and announcements section will grow and include some video testimonials.

The links to organizations will be live, adding IMDSA and the Jérôme Lejeune Foundation. They thanked **Mitchell Levitz, self-advocate, University Center for Excellence in Developmental Disabilities at the Westchester Institute for Human Development**, for beta testing, and NICHD has included their feedback. Most beta testers took about 10 to 15 minutes. NICHD has tried to streamline the online consent form; there are also assent options. Once they have set up an account, users can access “my profile.” The site also has a section for resources such as a glossary of medical terminology.

Two suggestions were given by DSC members based on Dr. Parisi and Ms. Miller’s presentation: (1) adding translocation mosaic as an option and (2) adding an “unsure” button, since some questions will not apply to everyone.

Dr. Maddox, NICHD, noted how helpful the Institutional Review Board had been. It had approved the site with some ongoing tweaks.

Dr. Melissa Parisi, NICHD, reported that NICHD has aggregated data to share and can look at responses to specific questions in total. It encourages participants to go to the “growth measurements” tab. There is the option to add attachments (e.g., genetic testing results). Once a person registers as a professional, you can see the aggregated data.

DSC members posed questions regarding the security of the site and how to advertise the launch. NICHD noted that information would be private and the site would be compliant with the Federal Information Security Management Act. NICHD is also working on a press release that could be provided to each organization. There was a plan to hold webinars and produce a how-to video. These outreach efforts

would be done in a staggered manner in order not to overwhelm the site. NICHD will continue to provide outreach materials, such as posters or flyers that can be downloaded. Consortium members expressed support and some noted that other groups with which they work would be happy to pass the information along.

Next Steps

Lisa Kaeser, NICHD, reported that NICHD plans to send out a press release Wednesday, but with language and talking points regarding upcoming webinars. NICHD will include language stating that the registry is “not international” for now.

Attendees

Sujata Bardhan, NICHD

Kyle Brown, Patient Crossroads (guest)

Marilyn Bull, American Academy of Pediatrics

Jon Colman, National Down Syndrome Society

Michael Harpold, Down Syndrome Research and Treatment Foundation

Brandy Hellard, IMDSA

Colleen Jackson-Cook, IMDSA

George Jesien, Association of University Centers on Disabilities

Lisa Kaeser, NICHD

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

Yvonne Maddox, NICHD

Mary Lou Oster-Granite, NICHD

Melissa Parisi, NICHD

Vanessa Rangel Miller, PatientCrossroads (guest)

Bob Riddle, National Institute of Neurological Disorders and Stroke

Laurie Ryan, National Institute on Aging

Robert Schoen, Research Down Syndrome

Charlene Schramm, National Heart, Lung, and Blood Institute

David Tolleson, National Down Syndrome Congress