

Inaugural Meeting of the Down Syndrome Consortium: *A Public-Private Partnership*

September 29, 2011

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

Purpose of Meeting

To hold the inaugural meeting of the Down Syndrome Consortium (DSC). Each person present represents a national organization interested in Down syndrome (DS) research and treatment and has agreed to commit to the purpose and endeavors of the DSC. A Memorandum of Understanding will be signed by representatives of each organization.

Welcome and Introductions

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

Dr. Maddox, Deputy Director of NICHD of the National Institutes of Health (NIH), informed the group of the purpose of forming the DSC. A press release announcing the formation of the DSC was to be released later that day, and photographs of the group would be taken for an article in the *NIH Record*. Web links to the press release and copies of any photographs for individual press communications were shared with the members of the DSC.

Background

Yvonne Maddox, NICHD

In December 2007, the NIH published a "NIH Research Plan for Down Syndrome." Among many other things, the plan called for the development of research resources for the DS field. In December 2010, the *National Conference on Patient Registries, Research Databases, and Biobanks* was convened. The DSC participants represent a public-private partnership designed to discuss current DS research and research needs, including the development and implementation of a registry. DSC participants were requested to announce their involvement in the DSC through notices or press releases and to mention the formation of the DSC at national and international meetings.

Goal: For the DSC to revolutionize how DS syndrome research is conducted, advance how partnerships are formed, and increase communication with stakeholders. The intent is to grow purposefully; however, the goal of the DSC is stated broadly to allow for a range of future activities.

Note: NIH does not routinely fund registries. However, it is understood that a national DS registry is critical to advancing clinical research. The DSC will provide input into this endeavor and others related to DS research.

Discussion

Discussion centered on process and communications. Dr. Maddox emphasized that NICHD is seeking support and ideas from the private partners to formulate and meet the goals of the DSC and create a sustainable vision and strategic plan.

It was noted that the DSC would serve as a two-way street. It can solicit input for NIH from private organizations and provide them with information and resources to inform their own work. It also could help to inform the community about the progress at NIH.

NIH Commitment Statement from the Trans-NIH Down Syndrome Working Group

Dr. Laurie Ryan, National Institute on Aging (NIA), spoke as the NIH representative to voice NIH's commitment to the DSC. She gave a brief presentation on a similar group within NIA called the Alzheimer's Disease Neuroimaging Initiative (ADNI) that is organized through a cooperative agreement. The ADNI has designed a longitudinal, natural history study that includes a range of participants from those experiencing the normal aging process to those suffering major cognitive impairment.

Dr. Ryan explained that the ADNI works through the NIH Foundation. She reviewed the ADNI's public-private network structure, which includes multiple NIH Institutes; private partners; and representatives from other government agencies, such as the Food and Drug Administration. Dr. Ryan described how this collaboration has allowed the ADNI to leverage resources and has spawned international efforts. She stated that the DSC is a great springboard for DS research and could lead to international collaborations.

NIH Commitment Statement from Private Partners

Dr. Michael Harpold, Down Syndrome Research and Treatment Foundation (DSRTF), spoke on behalf of private organizations participating in the DSC and emphasized their commitment to the DSC goals. He stressed its importance and reaffirmed the idea that this partnership can be a two-way street to help both public and private sectors committed to DS research. Dr. Harpold noted the strength of the diversity of perspective the DSC will have due to its members if they can arrive at a consensus and truly share or integrate existing networks and resources. He stated that international participation should be a goal.

NIH Overview and Discussion of the Draft Memorandum of Understanding

Susan Streufert and Lisa Kaeser created the draft Memorandum of Understanding. The document was designed to allow the DSC to be nimble in its activities (Appendix 1). NICHD will be responsible for coordination and travel. The intent is for DSC to meet two to three times per year in person or via webinar with an eye to cost and scheduling convenience.

NIH is creating a DS website that will serve NIH and the community at large. It will provide resources for researchers, clinicians, and families. The DSC may be called upon to consider content for this website.

The Memorandum of Understanding was given a date for signatures of Friday, October 7, 2011.

The Memorandum of Understanding was drafted to allow members to vote on decisions made by the DSC, which distinguishes it from a letter of agreement. The final document was to be provided to all DSC members within 24 hours of the meeting.

Dr. Maddox told the DSC that NIH pledges to maintain communication and transparency.

DSC members made suggestions for ad hoc members who had registry expertise.

Contact Registries for DS

Melissa Parisi, NICHD

Presentation attached

Dr. Parisi led the discussion about creating a national registry for individuals with DS and provided some general background information. She noted that there are three separate but linked resources: patient registries, research databases, and biobanks. Each serves a unique purpose, and everyone should be consistent when using this terminology:

- A *contact registry* is typically coordinated by an organization or researchers wherein individual registries. Information sharing sometimes takes place here.
- A *research database* is a research tool created by scientists or clinicians. It may be designed to answer specific research questions or to consider aspects related to longitudinal natural history studies.
- *Biobanks* are repositories of biological samples (e.g., tissue, organ, fluids).

In an ideal world, the three tools would be linked.

It was generally agreed that the content of the national contact registry should be short and include minimal clinical information, with a focus on contact information and the ability to indicate whether the registrant would be interested in being contacted about ongoing research.

Aspects to consider include the following:

- We must discuss how the registry should be structured (e.g., Web-based design, paper-based option), informed consent, governance (e.g., a review board as an intermediary, the review process for proposed research, Institutional Review Board [IRB] approval), and ownership and hosting of the registry.
- A global unique identifier (GUID) would be specific to each participant. GUIDs have been discussed in some research circles but are not yet common practice.
- Could it serve as a place to advertise clinical studies?
- Use of existing infrastructure could minimize startup and maintenance costs. Consideration of existing registries can help the DSC determine the preferred approach.
- Dr. Parisi reviewed several existing registries, such as the Global Rare Diseases Patient Registry and Data Repository and the Rare Disease Clinical Research Network Contact Registry. She noted that the template used by these networks could be modified for a DS registry. Another website, ResearchMatch.org, brings together researchers and patients and utilizes the platform of the Clinical Translational Science Awards (CTSA) consortium; this website is a partner to the CTSA databases called RedCap.
- Dr. Parisi provided an example of the content of contact registries. The DSC should help define the critical elements of information in the registry, such as NIH standard procedures of race and ethnicity, primary language, location of care, and the type of research in which the DSC is interested.

Note: A NICHD Request for Information soliciting information from the DS community about the need to develop a research database received more than 2,000 responses. The community was also queried about the need for a DS biobank.

Funding opportunities: Dr. Parisi briefly reviewed some of the current funding opportunity announcements in DS research, including “Understanding and Treating Comorbid Conditions in Adolescents with Intellectual and Developmental Disabilities” and “Preclinical Research on Model Organisms to Predict Treatment Outcomes for Disorders Associated with Attention Deficit Disorder.” She noted that a recently published article considered a working model to improve the lives of people with DS and that a partnership is required to see it through to fruition. The DSC could be that partnership.

Considerations: The DSC discussed a range of issues important to consider as the registry is established.

- Should a registry subcommittee be formed to handle this? Think of the pros and cons to designating a university to host the registry, Request for Proposal (RFP) options, funding, ownership, the visibility of the registry, and the focus of the registry subcommittee.
- How much time should the registry business plan cover — 5 years? Should we draft a two- or three-page plan?
- The review board should oversee the registry.
- For the protection of children, should we allow only basic information in the registry?
- Consider federal guidelines related to human subjects' protection changing.
- The U.S. Department of Health and Human Services would be considered the steward of the registry and research database, but it would not own the information.
- Individuals own their information, and they would be able to “opt out.”
- Will there be cooperation with other registries? How do we address the concern that the national registry will somehow diminish the existing regional registries?
- We need to define how to register consent, since it is not standardized.
- The registry could be designed to alert the staff when a child becomes of legal age (which would help with triggering awareness for need of legal guardianship of adults with DS).
- Consider the enrollment of DS families.
- Weigh the pros and cons of genetic information in a registry versus a biobank.

Hosting

- The registry shall be hosted on nongovernment property, such as an academic institution. Look to cost sharing, avoidance of fragmentation, accountability, and clearly defined roles of management and oversight.
- Will there be IRB oversight?
- Will there be an RFP for the hosting and cost of the registry?
- The registry website should be user friendly and robust but not overwhelming.

Marketing

- The DSC will have to market the registry carefully to families and maintain engagement. ADNI is a good example.

Funding

- One possibility is outside organization funding through the NIH Foundation. Alternatively, the DSC could be funded externally through private fundraising, and NIH could contribute in nonmonetary ways. Any money raised by private organizations would be donated to the NIH Foundation for use toward the registry. Will there be an RFP?
- If one university seems most able to house the registry, NIH could provide the funds.
- People outside NIH, such as some members of the DSC, are not allowed to partake in the funding review process. Proposals could be judged by a steering committee as opposed to the DSC.
- Be warned that it could be problematic if the funds for the registry are deeply entrenched in the government.

Action Items

1. The DSC agreed to form a subcommittee specifically for the DS national registry. Consultants could lend specific expertise. The subcommittee will develop a plan or several options to present to the DSC. The DSC will make the final decisions.
2. Dr. Maddox asked Dr. Parisi to serve as co-chair of the registry subcommittee; Dr. Parisi agreed.

3. Jon Colman, National Down Syndrome Society (NDSS), agreed to serve as co-chair. The co-chairs will determine the remaining members of the subcommittee, including any consultants with specific areas of expertise.
4. It was recommended that the informed-consent document dictate the maximum number of times per year that a participant would be contacted.
5. Recommend who will host the registry or how that will be determined (e.g., select individually, post an RFP).
6. Possible funding mechanisms, management, and oversight procedures will be prepared and discussed at the next meeting.
7. Recommend a funding mechanism.
8. Consider participant engagements and information and results sharing.
9. Consider information technology needs and maintenance.
10. Develop a 5-year business plan.
11. Develop a project management plan.

Next Steps and Concluding Remarks

The DSC discussed the timing for the next meeting. While some members believed that the registry subcommittee should be given sufficient time to make significant progress, others noted that the next meeting need not focus on the registry and that an update from the subcommittee would suffice.

Consideration and discussion was had surrounding dates and the many professional meetings already scheduled starting in February 2012. The next agenda should include the following:

- Revision of the DS strategic research plan. The original document was written in 2007; the 5-year review is in 2012, allowing time for consideration.
- Whether any agreed-upon metrics will be used to assess the performance of the previous research agenda.
- Questions that might be asked and responses before the plan's revision.

The meeting adjourned at 12:00 p.m. EDT.

Attendees

George Capone, Down Syndrome Medical Interest Group
Michelle Livingston, Global Down Syndrome Foundation
Jon Colman, NDSS
Yvonne Maddox, NICHD
Mashana Davis, NIH Library, Office of Research Services
Edward McCabe, Linda Crnic Institute for Down Syndrome
Lisa Gilotty, National Institute of Mental Health
Janelle Nanavati, Special Olympics International
Michael Harpold, DSRTF
Lynn Olson, American Academy of Pediatrics
George Jesien, Association of University Centers on Disabilities
Melissa Parisi, NICHD
Lisa Kaeser, NICHD
Bob Riddle, National Institute of Neurological Disorders and Stroke
James King, NIH Library, Office of Research Services
Laurie Ryan, NIA
Robert Schoen, Research Down Syndrome
Charlene Schramm, National Heart, Lung, and Blood Institute
Malcolm Smith, National Cancer Institute

Deanna Tharpe, Down Syndrome Affiliates in Action
David Tolleson, National Down Syndrome Congress

Appendix 1: Draft Memorandum of Understanding

MEMORANDUM OF UNDERSTANDING

among the

***EUNICE KENNEDY SHRIVER* NATIONAL INSTITUTE OF CHILD HEALTH AND HUMAN DEVELOPMENT**

Representing the NIH Down Syndrome Working Group

and

XXXXXXXXXXXXXXXXXXXX

establishing the

DOWN SYNDROME CONSORTIUM

The purpose of this Memorandum of Understanding (MOU) is to summarize the agreement among the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), _____, and _____ to establish the Down Syndrome Consortium, a forum for the purpose of exchanging information among representatives of various stakeholders in biomedical and behavioral research on Down syndrome (Ds).

Background

The National Institutes of Health (NIH) supports research on Ds through several of its component Institutes and Centers. Other agencies of the Federal government also address Ds research and policy, and many private groups and individuals are actively involved in some aspect of Ds research. In establishing this Consortium, the partner agencies and organizations agree that a single, comprehensive forum is needed to facilitate the exchange of information and to make the research effort to address Ds more efficient, effective, and transparent by assuring consistent communication, minimizing duplication of effort, and integrating the varied perspectives of the partner agencies, organizations, and individuals.

Purpose

The purpose of the Down Syndrome Consortium is to provide a forum for discussion regarding current research on Ds and continued implementation of the NIH Research Plan on Down Syndrome. Consortium members will represent a range of stakeholders committed to research on Ds, who will be encouraged to contribute opinions and views of research needs and opportunities from their unique perspectives.

Membership

The Consortium will include the following members:

- Members of the NIH Down Syndrome Working Group
- A self-advocate with Ds
- A family member of a person with Ds
- American Academy of Pediatrics
- American Association on Intellectual and Developmental Disabilities
- Association of University Centers on Disabilities
- Down Syndrome Affiliates in Action
- Down Syndrome Research and Treatment Foundation
- Down Syndrome Medical Interest Group
- Global Down Syndrome Foundation/Linda Crnic Institute

- National Down Syndrome Congress
- National Down Syndrome Society
- Research Down Syndrome
- Special Olympics
- *Ex officio* members as appropriate

Each organizational member will appoint a representative to participate in Consortium activities. Additional agencies, organizations or individuals may be invited by the full Consortium to join the group in the future, or to attend specific meetings, as needed, for the purpose of making presentations, sharing information, and/or discussing topics pertinent to the meeting agenda. Permanent additions to Consortium membership will be reflected in an addendum to this MOU.

Consortium Activities

Consortium members may, but are not limited to:

- Identify and agree upon the goals, procedures, and practices of the Consortium;
- Share information, data, and program activities related to Ds;
- Discuss research needs and priorities;
- Provide input on the method and timing of implementing components of the NIH Research Plan on Down syndrome, and provide periodic reviews and updates to the Plan as needed;
- Foster the effective collaboration of the partners on joint projects, as appropriate;
- Provide feedback on draft materials and products, as appropriate;
- Involve other Federal or non-Federal partners, as appropriate.

The Consortium will function as a public-private working group, with shared goals. It will not be advisory to the NIH. All members, individually or jointly, may be guided by the opinions expressed by the Consortium.

Roles and Responsibilities

The *Eunice Kennedy Shriver* National Institute of Child Health and Human Development will serve as the chair of the Consortium and will be responsible for scheduling and facilitating Consortium meetings and other communications, planning meeting logistics and recording minutes, and overseeing Consortium activities.

Together, the Consortium members will commit to attending meetings, fully participating in activities, bringing up current issues for discussion, completing action items as agreed upon, and reporting on relevant activities of their agency or organization.

Communications

Meetings of the Consortium will take place at least twice per year, either in person or via the Internet. Interim communications via telephone, email or other means will take place as needed.

Recordkeeping

A summary of Consortium meetings will be prepared within two weeks after each meeting, and distributed to Consortium members.

Funding

Nothing in this agreement shall be deemed to be a commitment or obligation of Federal or non-Federal personnel, funds or other resources. Activities described herein are contingent upon the

availability of funds for this purpose. This MOU and all associated agreements will be subject to the applicable policies, rules, regulations, and statutes under which the NIH operates.

Member Contacts

Eunice Kennedy Shriver National Institute of Child Health and Human Development:

(Mary Lou Oster-Granite or Melissa Parisi)

Contacts for each Consortium member

Duration of the Agreement

This agreement is at-will and may be modified by mutual written consent of authorized officials from the consortium members. This agreement shall become effective upon signature by authorized officials from the consortium members and will remain in effect until modified or terminated by all parties by mutual written consent. Individual membership may be terminated by any party upon thirty (30) days prior written notice to the other members.

Concurrence