



Down Syndrome Research and Advocacy: Improving Lives Together

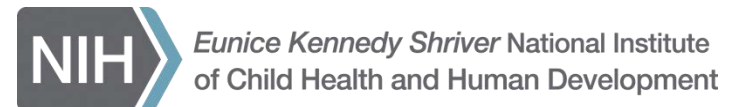
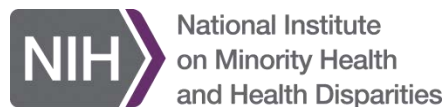
NACHHD Advisory Council Meeting ♦ June 5, 2014

Yvonne T. Maddox, Ph.D.

Acting Director

National Institute on Minority Health and Health Disparities (NIMHD)

National Institutes of Health (NIH)





Objectives

- ❖ Delineate some of the new research directions, opportunities, and challenges
- ❖ Discuss how public-private partnerships are mobilizing a global Down syndrome agenda
- ❖ Describe how advocacy has advanced research and future needs
- ❖ Provide an update on DS-Connect™: The Down Syndrome Registry



Introduction

- ❖ In recent years, basic science and animal studies have led us to a new vision for Down syndrome research.
- ❖ Partnerships from many sectors—government, academia, industry, advocacy organizations—are playing a critical role.
- ❖ While research is moving forward, there are challenges for researchers, clinical investigators, and the Down syndrome community.
- ❖ “Champions for Down Syndrome Research” are learning how they can share in overcoming these challenges.



Some research benefits for people with Down syndrome

- ❖ The advent of antibiotics, heart surgery, and specialized medical care have improved early survival and longevity for most people with Down syndrome.
- ❖ Medical and educational advances have depended on scientific evidence and social change.
- ❖ Evidence-based therapies and educational approaches are helping young people with Down syndrome today achieve more than ever before.



Recent advances in Down syndrome research

- ❖ Researchers have identified new compounds that may act on the brain in ways which could improve mental function (e.g., GABA receptor antagonists, beta adrenergic receptor agonists).
- ❖ Several mouse models for Down syndrome are available.
- ❖ Early phase 2 pharmaceutical compounds to improve learning and memory.



Challenges: conducting cognition research

- ❖ Ts65Dn mice are not humans with Down syndrome; treatments in mice may not have same effects in people.
- ❖ Some pharmaceutical therapies evaluated in humans have not been very encouraging, but we are hopeful about new therapies.
- ❖ We need to move forward cautiously; pharmaceuticals that may improve cognition, reduce dementia, and affect other nervous system and brain functions could have serious side effects.
- ❖ We need to evaluate all potential therapies in humans.



Importance of the Down syndrome community in biomedical research

- ❖ Families, self-advocates, and caregivers must encourage participation in clinical trials.
- ❖ Professionals have a responsibility to provide clear, accurate information and to be cautious about stating the eventual benefits that a trial may bring if there is no supporting data.
- ❖ Advances require a team effort with researchers, clinicians, people with Down syndrome, and their families working together.



Down Syndrome Consortium



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



National Cancer Institute (NCI)



National Heart, Lung and Blood Institute (NHLBI)

DOWN SYNDROME AFFILIATES IN ACTION



National Institute of Mental Health (NIMH)



National Institute of Neurological Disorders and Stroke (NINDS)



National Institute on Aging (NIA)




Special Olympics





DS-Connect™ Home Page



National Institutes of Health
Turning Discovery Into Health

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DS-Connect™: The Down Syndrome Registry

“The new registry provides an important resource to individuals with Down syndrome and their families,” said Yvonne T. Maddox, deputy director of the NIH’s *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), which is funding the registry. “The registry links those seeking volunteers for their research studies with those who most stand to benefit from the research.”

[Join the Registry](#)

Launched September 6, 2013



DS-Connect™: The Down Syndrome Registry

- ❖ Idea for a registry emerged from two 2010 meetings identifying important research resources.
- ❖ Registry is supported by the Down Syndrome Consortium membership.
- ❖ Contract awarded in September 2012 to Patient Crossroads.
- ❖ Purpose of the registry is to facilitate research.
- ❖ Tapping into the collective voice of individuals and families will improve our understanding about the condition and enhance research participation.

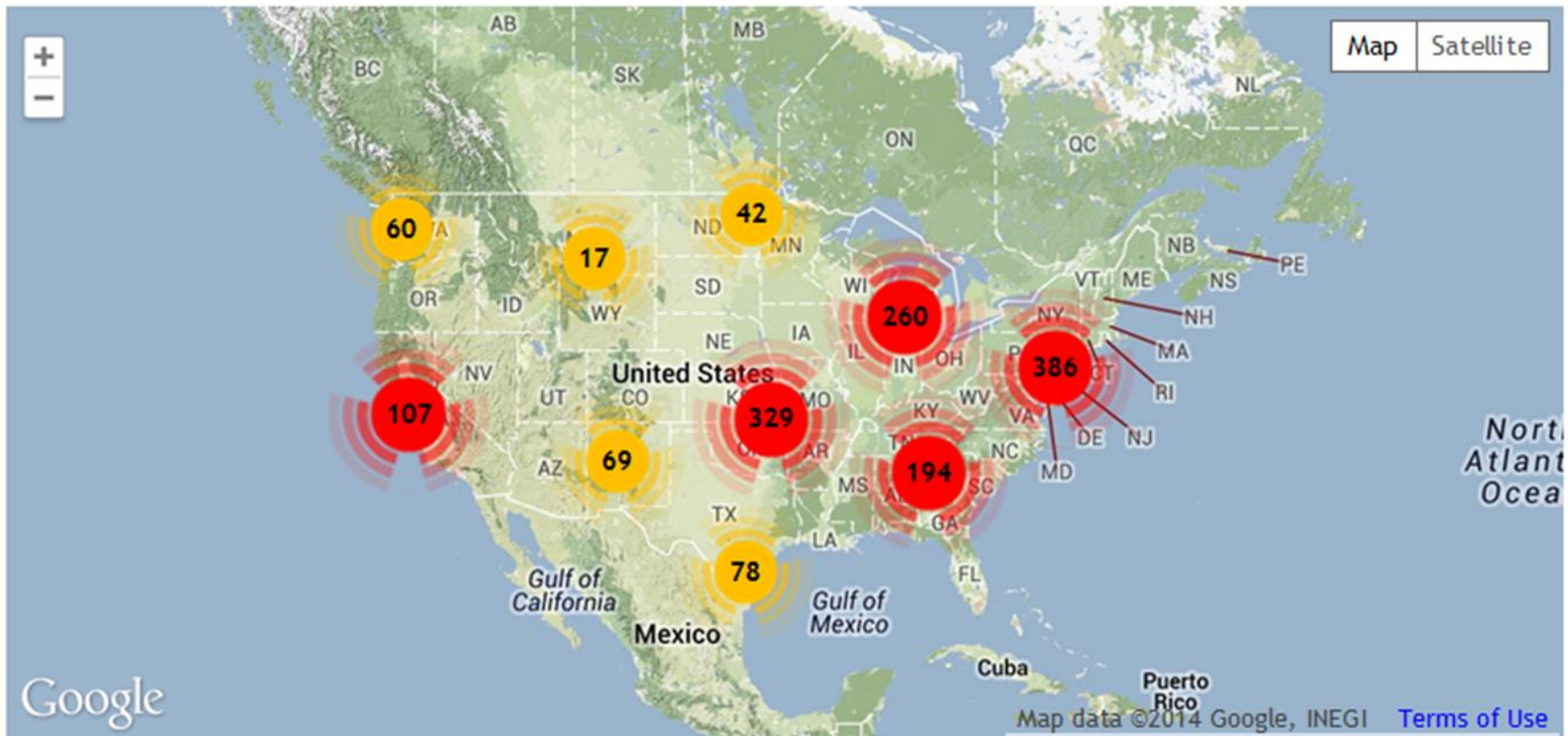


DS-Connect™: The Down Syndrome Registry

- ❖ Contact information and health history can be entered in an online, secure, confidential database.
- ❖ Registry participants can customize their profile, update it online, and choose the information they would like to share.
- ❖ Professional Portal: Clinicians and researchers who are authorized to access the database can contact the Registry Coordinator, with permission from participants, about recruitment in appropriate research studies.
- ❖ The Registry will become a clinical trials recruitment resource and evaluation tool.



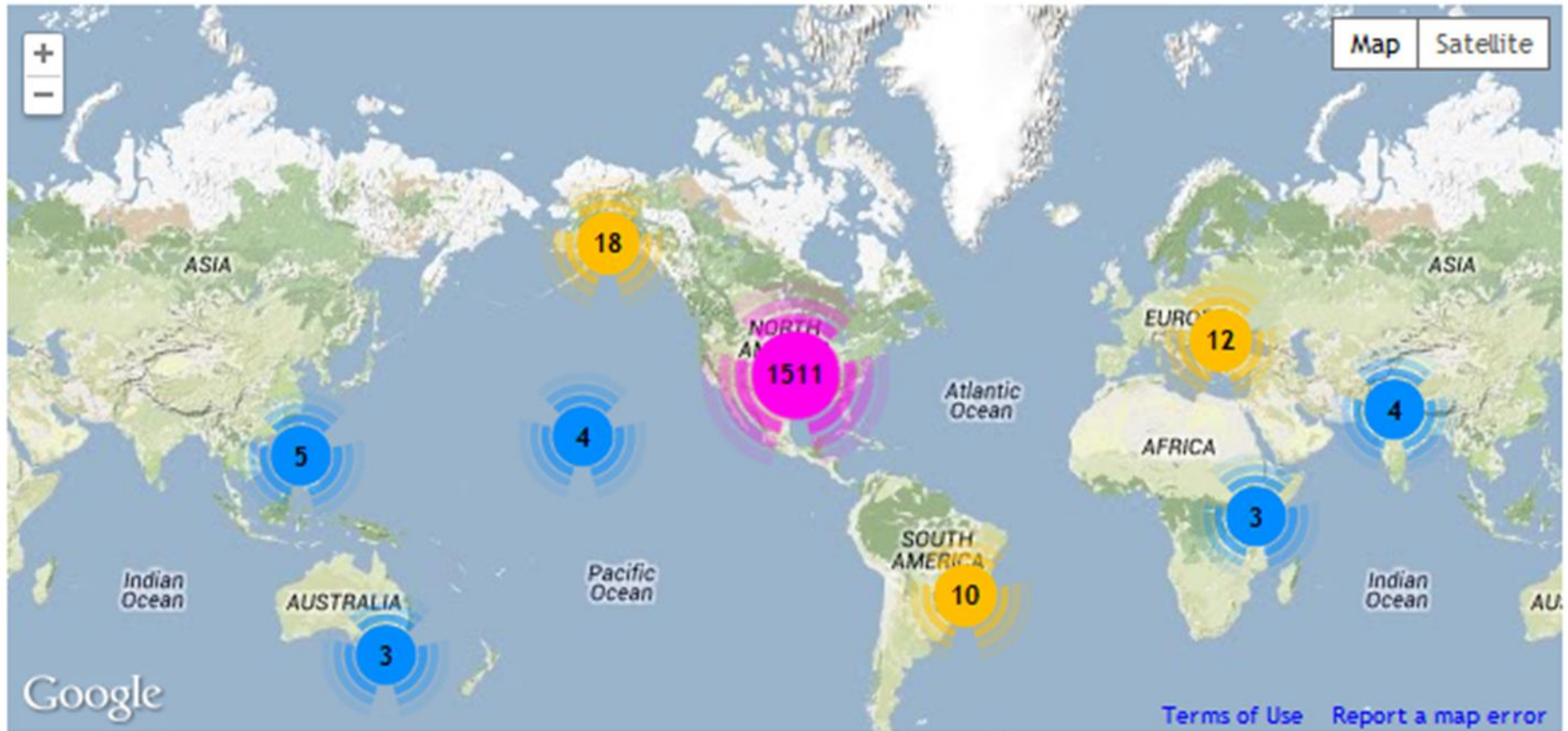
Current DS-Connect™ Registrants: North America



1,629 as of June 2, 2014



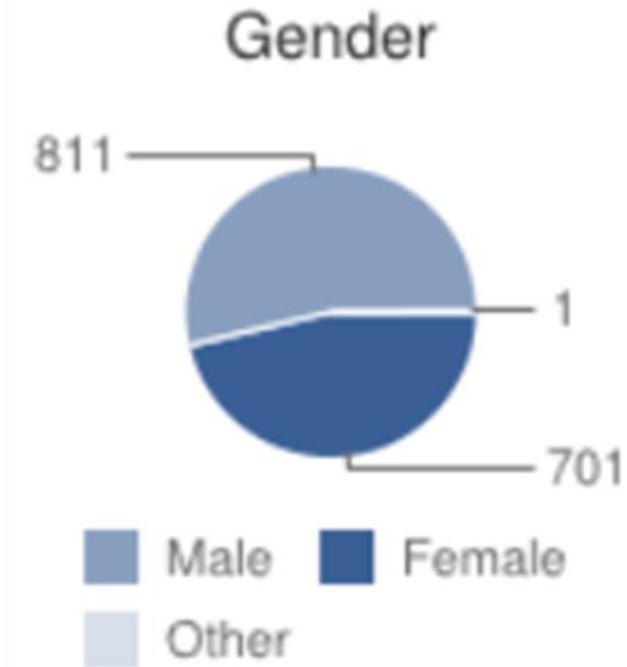
Current DS-Connect™ Registrants: Worldwide



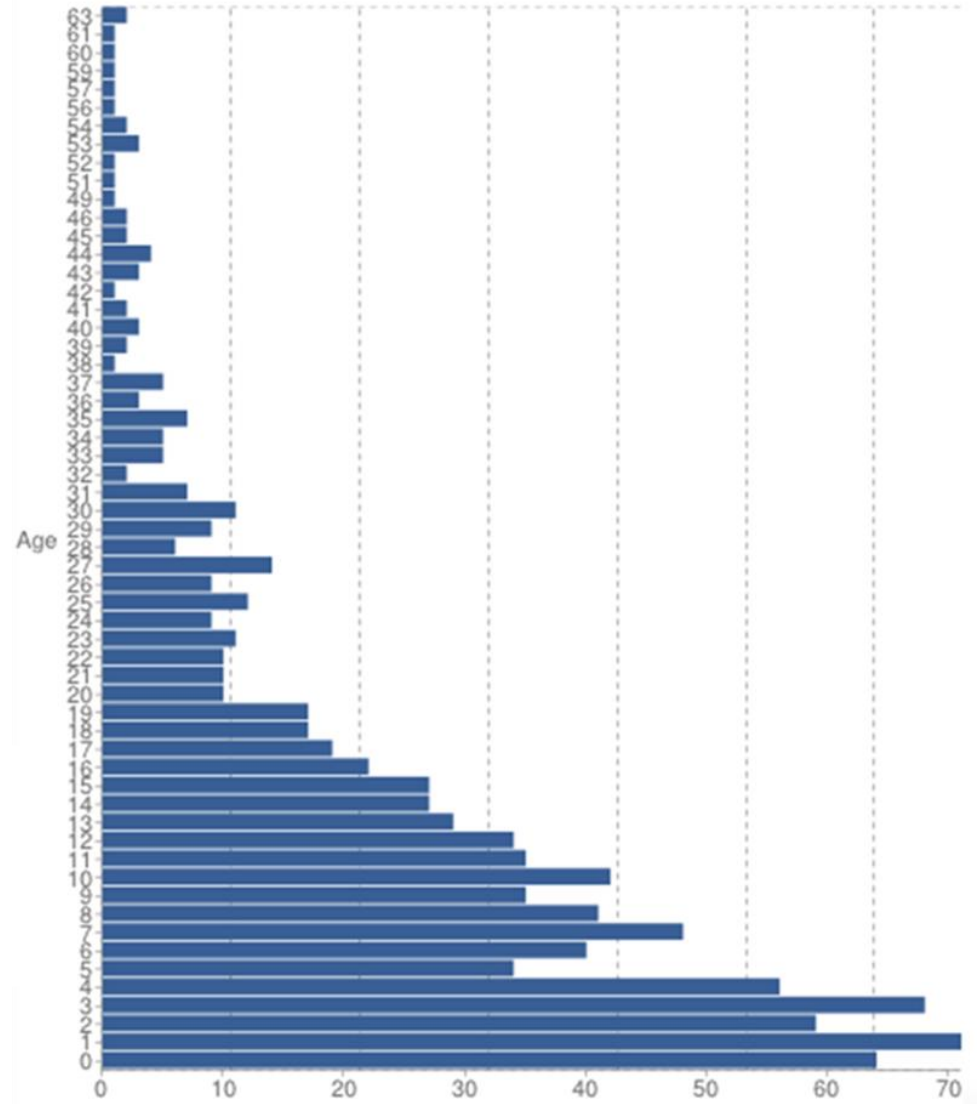
50 as of June 2, 2014



Explore the data: demographics



Under Age 30: 1286
Over Age 30: 108





How secure is it?

- ❖ Meets stringent data security requirements to protect personally identifiable information
 - ~ 250 security controls are checked regularly
 - Information is encrypted
 - Password requirements (At least 6 characters long, 1 upper case, 1 lower case, 1 symbol, 1 number)
Example: **P@ssword3**
 - Passwords must be changed every 6 months
 - No social security info is collected
 - No personal bank account info is collected






Portal for Professionals




Registry Updates




Get information on the latest registry-related news, including ongoing efforts to increase the quality of life of people living with Down syndrome and advance research focused on developing treatments. Access information on how to better manage symptoms, connect with online support resources, and more.

Professionals



Researchers, clinicians and other professionals can access de-identified aggregate data to identify populations for trial/study planning or recruitment. You can also contact DSPR to distribute study opportunities and notices to participants. [Register as a Professional](#) to get started.

Explore the Data



Check out how to explore the information you and others have provided to learn more about Down syndrome and its impact on other community members. Log into your account today and start exploring the data.

← Link available on home page

- ❖ No direct access to Registry participants
- ❖ Registry Coordinator will contact eligible families about the study or trial
- ❖ Participants can choose whether to contact investigators to join



Under
Development

Professional Portal: Tiers of access

- ❖ Access Level 1: View data identical to Registrants' view and can perform more detailed searches of de-identified data
- ❖ Access Level 2: More complex searches, statistical analysis
- ❖ Access Level 3: For study recruitment

NOTE: Investigators will **NEVER** be able to contact participants directly. The Registry Coordinator will inform the participant of any clinical study of interest.



Data Access Policy



❖ Data Access Policy:

- Review criteria for approving studies requesting specialized searches and study recruitment
- Develop publication/presentation guidelines
- Develop guidelines for industry studies

❖ A Data Access Committee will review these requests

- Members to include: Registry developers, NIH staff, researchers, families, other DS advocates



Future Developments

- ❖ Other Survey Modules: leukemia, female health, cause of death, glossary

- ❖ Spanish language version—launch in fall 2014

- ❖ Expanded resources section:
 - ClinicalTrials.gov and About Clinical Trials
 - Healthychildren.org information for families by AAP

- ❖ Customizable functions on participant account:
 - Educational component
 - Health care reminders
 - Messages about the Registry

- ❖ Link to biospecimen repositories via GUID



Giving Back to the DS Community

- ❖ For families:
- ❖ Lists of healthcare providers
- ❖ Print out survey answers and share them with medical providers
- ❖ Update DS growth charts
- ❖ NIH and scientists can know about health needs of individuals with DS of all ages
- ❖ Connect with researchers for future clinical studies
- ❖ **It only takes a few minutes...**

DS-Connect™

The Down Syndrome
Registry

<https://DSConnect.nih.gov>





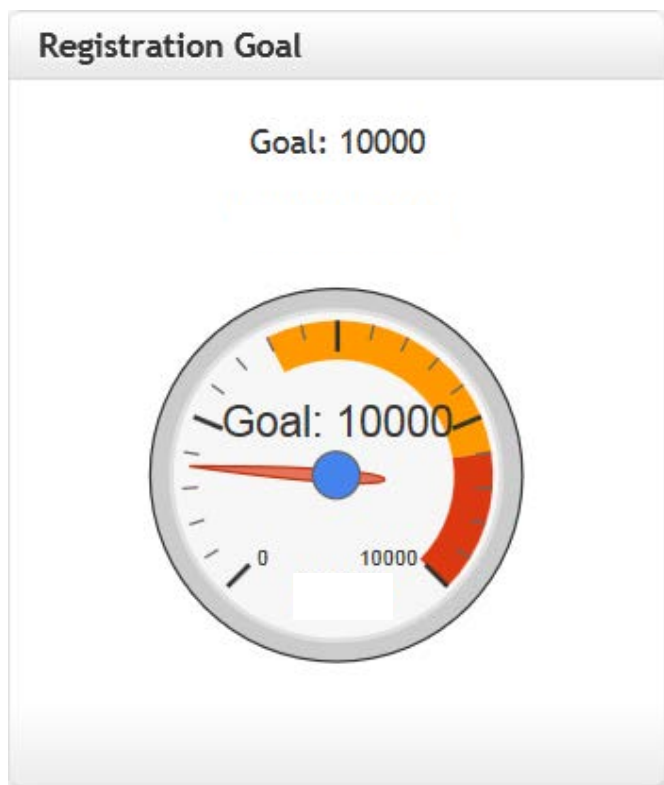
NIH: Goals for Down syndrome research

- ❖ Immediate goals:
 - Expand the DS-Connect Registry
 - Launch the Professional Portal
 - Update the NIH Research Plan on Down Syndrome
 - Support more clinical trials



NIH: Goals for Down syndrome research

- ❖ Ongoing goals:
 - Involve more families, clinicians, and scientists in Down syndrome research
 - Use research to inform parents and families about latest research-supported facts
 - Improve the lives of children, adolescents, and adults living with Down syndrome





Get DS Connected!

Share her health information—
help others.



Join DS-Connect™: The Down Syndrome Registry today.
Get DS-Connected: dconnect.nih.gov



Eunice Kennedy Shriver National Institute
of Child Health and Human Development

His health information matters.



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Eunice Kennedy Shriver National Institute
of Child Health and Human Development

<https://DSConnect.nih.gov>



The Goal: Helping all individuals reach their full potential.

<http://downsyndrome.nih.gov>

<http://DSCConnect.nih.gov>