



Getting DS-Connected: A new national registry for people with Down Syndrome

The Arc of Montgomery County ♦ June 11, 2014

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National Institutes of Health (NIH)



Eunice Kennedy Shriver National Institute
of Child Health and Human Development



Objectives

- ❖ Why is research important?
- ❖ What is DS-Connect™: The Down Syndrome Registry?
- ❖ Demonstration of the registry data
- ❖ FAQs
- ❖ Down syndrome research plan



Why should I care about research?

- ❖ Basic science and animal studies have led us to a new vision for Down syndrome research.
- ❖ Partnerships from many sectors—government, universities, 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, especially for cognition research.
- ❖ Recent advances in Down syndrome research have focused on early phase 2 medications to improve learning and memory.
- ❖ Advances require a team effort with researchers, clinicians, people with Down syndrome, and their families working together.



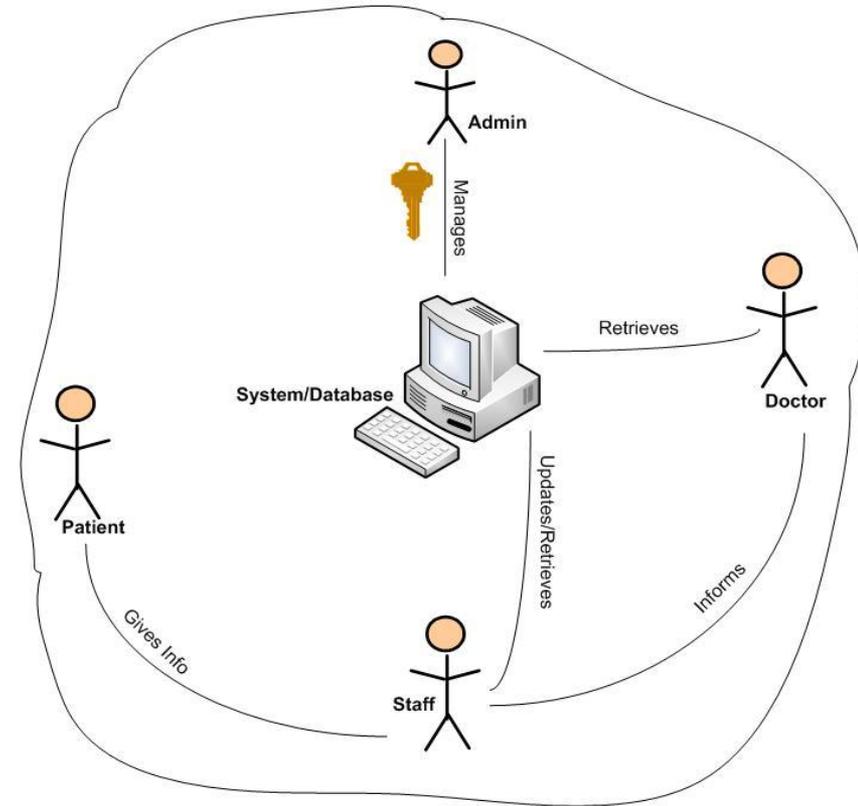
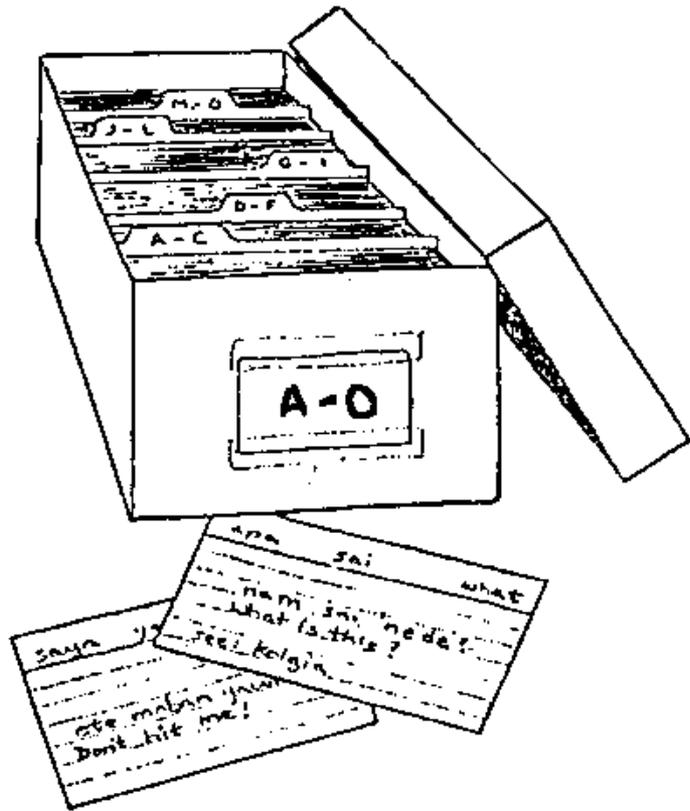
Goals of a Registry

- ❖ Collection of information about individuals with a specific diagnosis or condition
- ❖ Purposes:
 - Understand a condition and its “natural history”
 - Identify which medications or treatments are being used
 - Help scientists identify research projects to develop new treatments
 - Determine if an FDA-approved medication is safe or to meet other regulatory requirements (post-marketing surveillance)

Register
Today



Different types of registries





**Who knows
them best?**





YOU!!





A new model: Family-centered registries



- 1 **Foundations or institutions** sponsor registries, act as the trusted gatekeeper.
- 2 **Families or Self Advocates** register and provide detailed health history information.
- 3 **Coordinators** may curate data, answer questions, assist with participation.
- 4 **Clinicians/Researchers** have access to aggregated, de-identified data.
- 5 **Industry** uses the de-identified data for study planning and pre-screening for enrollment.



But that's not all...

- ❖ Document growth charts
- ❖ Access the healthcare provider list
- ❖ Print out my child's medical history
- ❖ Get access to healthcare guidelines
- ❖ Find out about research that impacts people with Down syndrome
- ❖ Connect with resources on Down syndrome





Why are we doing this?

- ❖ We had the participation and support of the DS advocacy and medical groups
- ❖ New clinical trial opportunities need willing volunteers
- ❖ We set aside the resources to create it
- ❖ No national DS registry existed, and families asked for one!



A Brief History of DS-Connect™

- ❖ 2010: Meetings were held to discuss a registry...
- ❖ 2011: The DS Consortium was founded by NIH
- ❖ 2012: A contract was awarded to PatientCrossroads
- ❖ 2013: DS-Connect was launched
- ❖ 2014: Professional Portal will be launched



DS-Connect™

What it is:

A secure, confidential, online survey tool to collect basic information about people with Down syndrome

Goals of DS-Connect™:

- ❖ To identify the features of DS across a large population of those with DS
- ❖ To identify individuals with DS who may be eligible for research studies
- ❖ To allow individuals with DS and their families to look at their information and compare it to de-identified information from other Registry participants

Features:

- ❖ Potential to aggregate data from individuals with DS globally
- ❖ Provide a database and educational system that has value for those with DS
- ❖ Enable researchers to use de-identified data to develop studies on etiology, natural history, and treatments



How does it work?

- ❖ The **Down syndrome Consortium**: supports and publicizes the Registry
- ❖ The **Registry Team**: manages the content and sends out notices and other information.
- ❖ Two oversight boards for DS-Connect™
 - The **DS-Connect Operations Board** oversees the day-to-day activities of the Registry, develops surveys, and makes decisions about access to Registry information. It also works with the registry coordinators and Governance Board to implement policies.
 - The **DS-Connect Governance Board** makes decisions about the overall policies and procedures that govern the Registry.



DS-Connect: Who is involved?

DS 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)





DS-Connect: Who is involved?

Registry Team

- Melissa Parisi, M.D., Ph.D.
Chief, Intellectual and Developmental Disabilities Branch, NICHD
- Lisa Kaeser, J.D.
Director, Office of Legislation and Policy, NICHD
- Aubrey Callwood
Information System Security Officer, NICHD
- Sujata Bardhan, Ph.D.
Scientific Program Manager
Intellectual and Developmental Disabilities Branch, NICHD
- Debbie Jae, M.S.
Program Director, PatientCrossroads
- Albert Carrasco
Systems Engineer, PatientCrossroads



DS-Connect: Who is involved?

Operations Board

- Peter Bulova, M.D.
University of Pittsburgh
- Priya Kishnani, M.D., MBBS
Duke University School of Medicine
- Stephanie Sherman, Ph.D.
Emory University
- Wayne Silverman, Ph.D.
Kennedy Krieger Institute
- Sujata Bardhan, Ph.D., NICHD
- Michael Bender, Ph.D., NIGMS
- Mary Lou Oster-Granite, Ph.D., NICHD
- Melissa Parisi, M.D., Ph.D., NICHD
- Robert Riddle, Ph.D., NINDS
- Yaffa Rubinstein, Ph.D., NCATS
- Laurie Ryan, Ph.D., NIA



DS-Connect: Who is involved?

Governance Board

- Mark L. Batshaw, M.D.
Children's National Medical Center
- Tom Blumenthal, Ph.D.
Linda Crnic Institute for DS
- Marilyn J. Bull, M.D.
American Academy of Pediatrics
- Elisabeth Dykens, Ph.D.
Vanderbilt Kennedy Center
- Gary Goldstein, M.D.
Kennedy Krieger Institute
- Michael M. Harpold, Ph.D.
LuMind Foundation
- Claire Henkhaus
Parent
- Lisa Kaeser, J.D.
NICHD
- John Mathewson
HSC Foundation
- David Tolleson
National Down Syndrome Congress



DS-Connect™ Home Page



National Institutes of Health
Turning Discovery Into Health

[Forgot login?](#)

[Need Help?](#)

- Home
- Contact Us
- About DS-Connect™
- News
- Resources
- For Professionals

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



Registration page for individuals and families

Steps for participating in the Registry

There are four easy steps to join DS-Connect™. This whole process should take you less than 20 minutes. If you are a self advocate, you may want to have a trusted person help you with these steps.

1. Create an account: The account can be created by choosing a user name and a password. This will be used each time you want to log in to your profile.
2. Review the online consent form.
3. Click the appropriate consent boxes, enter the security code, and click “Register.”
4. Once you are registered, you can enter basic information for the participant with Down syndrome (DS). If you are a person with DS, you may want to have someone you trust help you to join the registry.

Step 1: Create an Account

Your First Name:	<input type="text"/>	
Your Last Name:	<input type="text"/>	
Please select the best option:		
Your Relationship to Participant with DS:	<input type="text" value="v"/>	 
Your Email / Re-enter email:	<input type="text"/>	 
Secondary Email:	<input type="text"/>	
Your Username:	<input type="text"/>	 
Your Password / Re-enter password:	<input type="text"/>	 



DS-Connect™: Provide Consent

Step 3: Click the appropriate consent boxes, enter the security code, and click “Register.”

For the purpose of this document “I” and “my” refers to the registrant, either the person with DS or the parent, guardian or legally authorized representative providing the information on behalf of the person with DS (The legally authorized representative is the person legally responsible for the care of the person with DS).

-  I understand the purpose and nature of DS-Connect™, and I have no additional questions.
-  My participation in this project is entirely my choice. If I change my mind and wish to withdraw my information from DS-Connect™, I will be free to do so without having to provide any explanation. I understand this.
-  I understand the risks and benefits of participation and I agree to participate in DS-Connect™.



DS-Connect™: Choose Consent Options

Consent Options

Please choose the consent that applies to you

I have read the explanation about DS-Connect™ and have been given the opportunity to discuss it and ask questions. I hereby consent, or give permission for my child or the participant with DS to take part in this registry.

- Consent by the adult participant with DS (if the person with DS can provide their own consent)
- Permission by a Legal Guardian or Legally Authorized Representative (if the person with DS is 18 years or older and cannot provide their own consent)
- Parent's permission for minor (if the person with DS is under 18 years of age)





DS-Connect™: Choose Assent Options

Please choose the assent option that applies to the person with DS:

We request assent if the person with DS is 7 years of age or older and is unable to provide consent but is able to give assent to participate.

- Yes, the information in the above consent was described to the participant with DS who is 7 years of age or older and he/she agrees to participate in DS-Connect™.
- Not applicable. The participant with DS is not able to provide assent.
- Not applicable. The participant with DS is over 18 years of age and has provided his/her informed consent as above.



Please enter the code below



Register

 Required field |  Information: Point mouse to icon



Sharing and Contact Preferences

Sharing and Contact Preferences

 DS-Connect™ has processes in place to protect your identity, including saving your information with a code instead of your name so others don't know who you are. DS-Connect™ may share information with scientists for research studies or clinical trials, but it will not share your personal information—see the explanation of “de-identified” above. **Do you give your permission for DS-Connect™ to share your de-identified information for research studies or clinical trials?**

(Please note that even if the coordinators of a clinical trial believe that you might be eligible for the trial based on your data in the Registry, it is still possible that you will not meet the trial inclusion criteria after all. Please also note that if we inform you about the existence of a trial, it does not mean that we endorse or support it. In order to take part in any trial, you will need to fill out a separate informed consent form.)

 DS-Connect™ may share your de-identified information with other registries or databases. **Do you give your permission for DS-Connect™ to share your de-identified information with other registries and databases?**

 DS-Connect™ may get information about a study to donate samples of blood, tissue or other biospecimens for research. **Do you want DS-Connect™ to contact you about these types of studies?**

 If researchers learn anything interesting about your condition, **do you want DS-Connect™ to contact you with this information?**

 It is important that the DS-Connect™ information is up to date. We will contact you once a year to ask about changes in your health condition. **Do you give us permission to contact you for this information?**



Sample Question

Initial Health Questionnaire - Health History

Progress

16%

Has the participant ever had any cardiac or heart surgery? (Select all that apply.)

- None
- Yes, surgery for a congenital heart defect in the first year of life
- Yes, surgery for a congenital heart defect after the first year of life
- Cardiac surgery for other reason later in life
- Unsure



Sample Trigger Question

Initial Health Questionnaire - Endocrine

Progress

42%

Has the participant ever been diagnosed with a thyroid condition?*

No

Yes

Unsure



Multiple Survey Modules

Initial Health Survey with “Trigger Questions” that lead to other surveys:



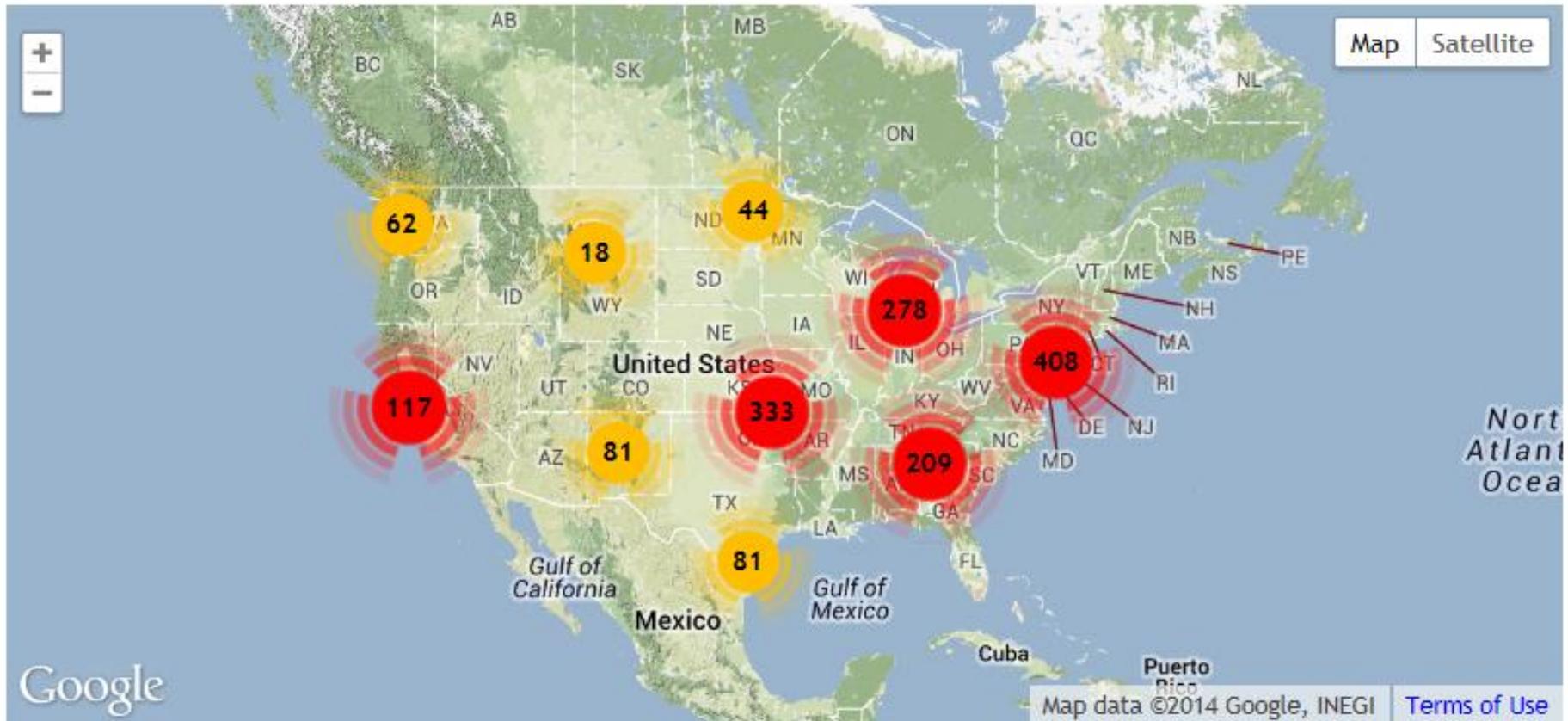
- Initial Health Questionnaire
- Thyroid Questionnaire
- Heart Questionnaire
- Sleep Questionnaire
- Skeletal Questionnaire
- Gastrointestinal Questionnaire
- Adulthood Questionnaire
- Diabetes Questionnaire
- Celiac Disease Questionnaire
- Development Questionnaire
- Prenatal and Birth Questionnaire

Available to all registrants





Current DS-Connect™ Registrants: North America



1,622 as of June 4, 2014



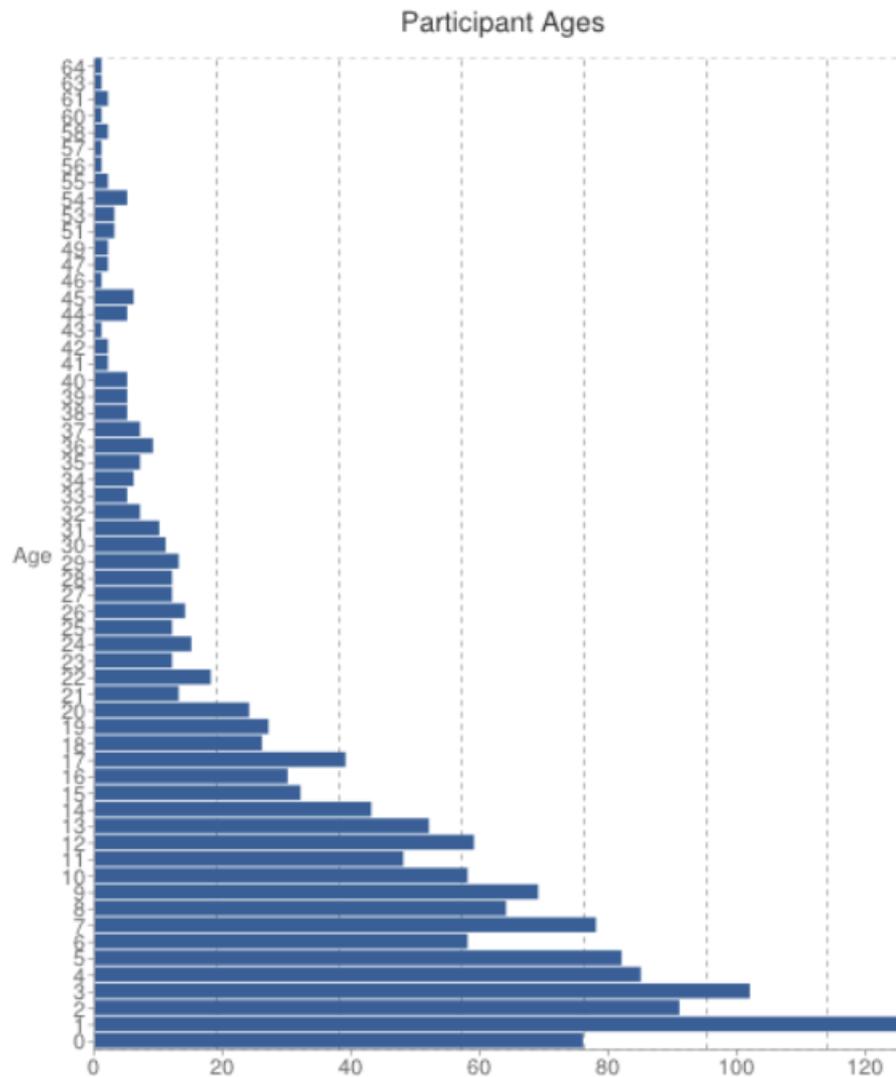
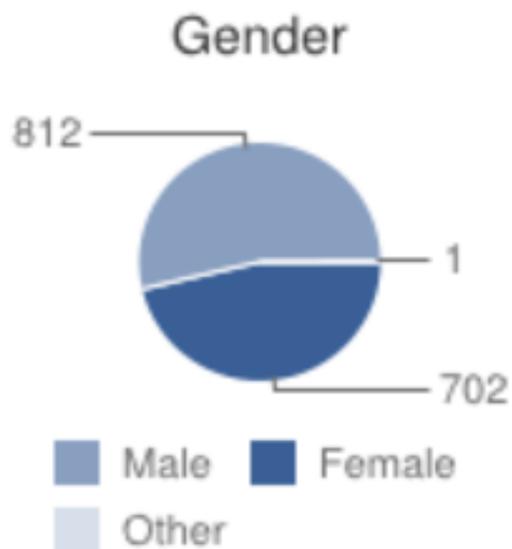
Registrants World-wide



59 outside U.S. as of June 4, 2014



Explore the data: demographics



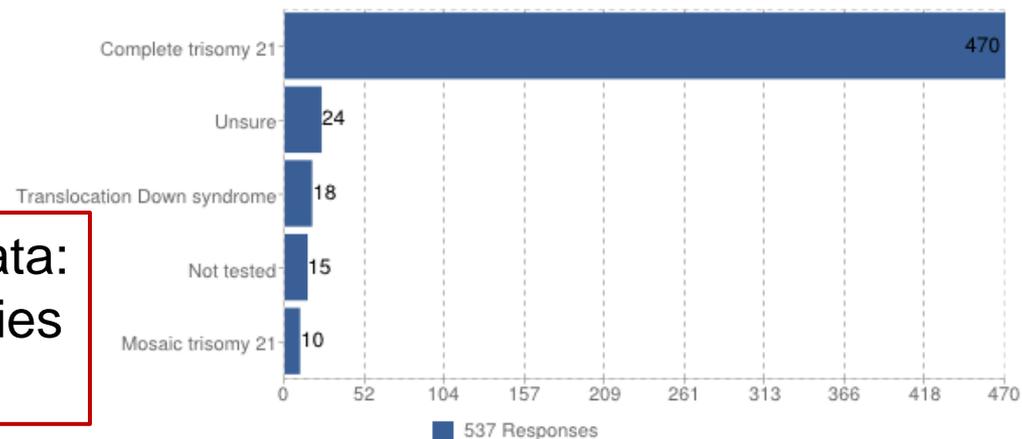


Explore the Data

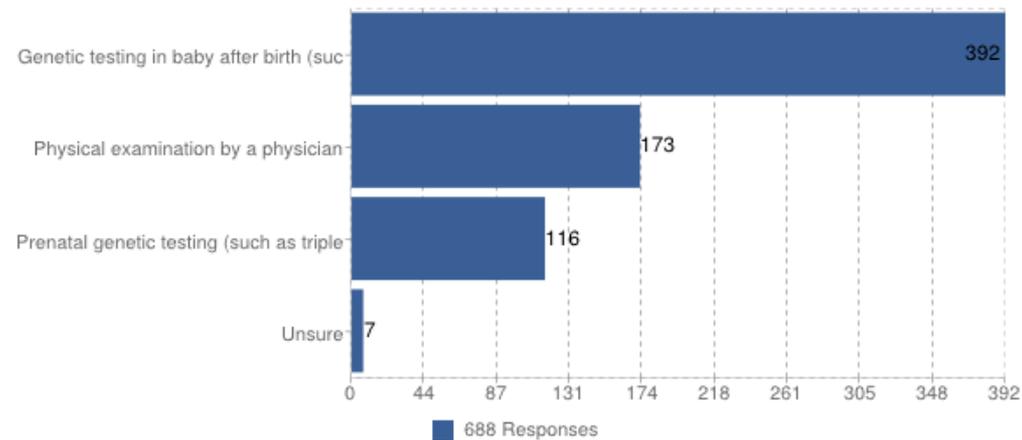
Aggregated, de-identified data:
Available to registered families
and professionals

Initial Health Questionnaire - Diagnosis

What is the participant's Down syndrome diagnosis?



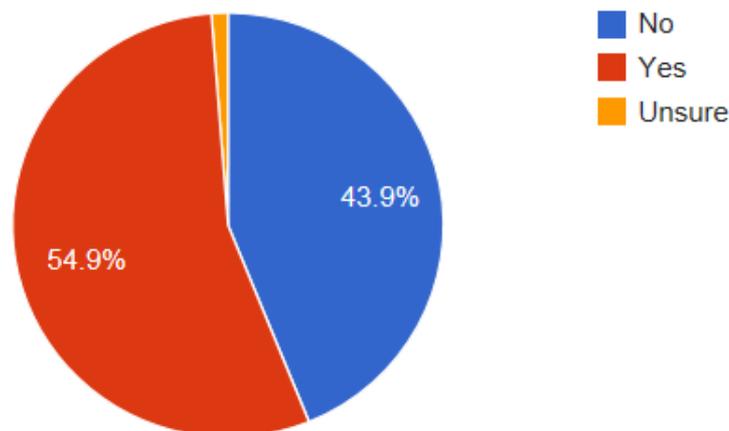
How was the diagnosis of Down syndrome made? (Select all that apply.)



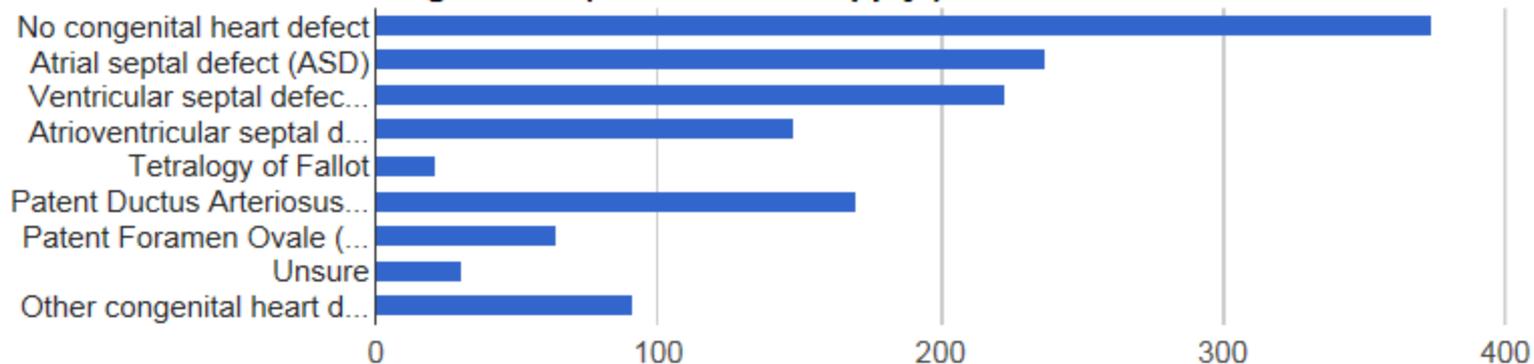


Explore the Data: Heart Defects

Has the participant been diagnosed with a congenital heart defect
(a birth defect of the heart)? (1133 responses)



Which of the following congenital heart defects have been
diagnosed? (Select all that apply.)

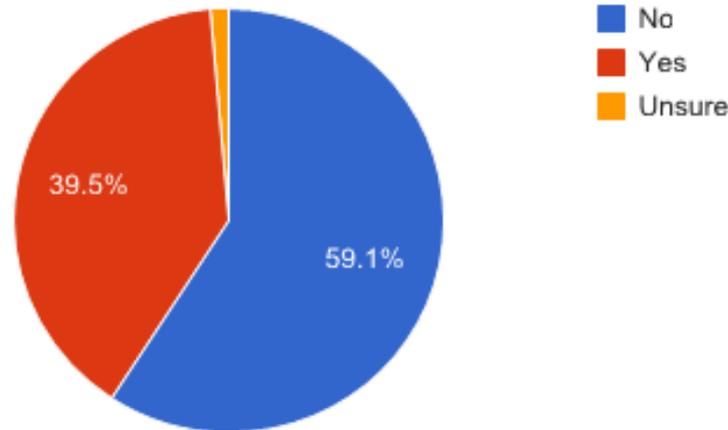


1014 people provided 1358 responses.

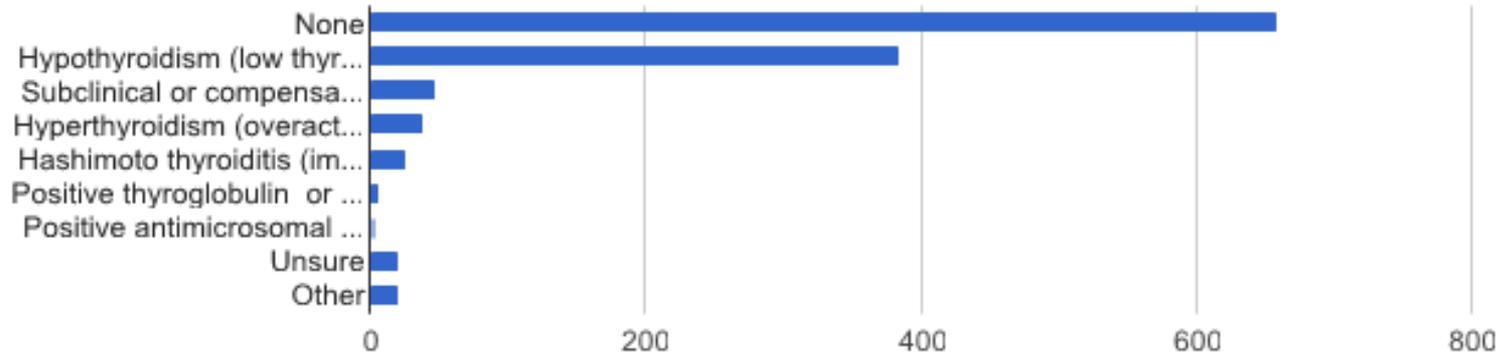


Explore the Data: Thyroid

Has the participant ever been diagnosed with a thyroid condition? (1226 responses)



What types of thyroid condition(s) have been diagnosed?
(Select all that apply.)



1160 people provided 1211 responses.



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



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.



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



Electronic Tool-kit

❖ Resources available:

- Facebook info-cards
- Badges
- Tweets
- Flyers

❖ All resources free of charge

❖ Contact us at:

DSConnect@nih.gov

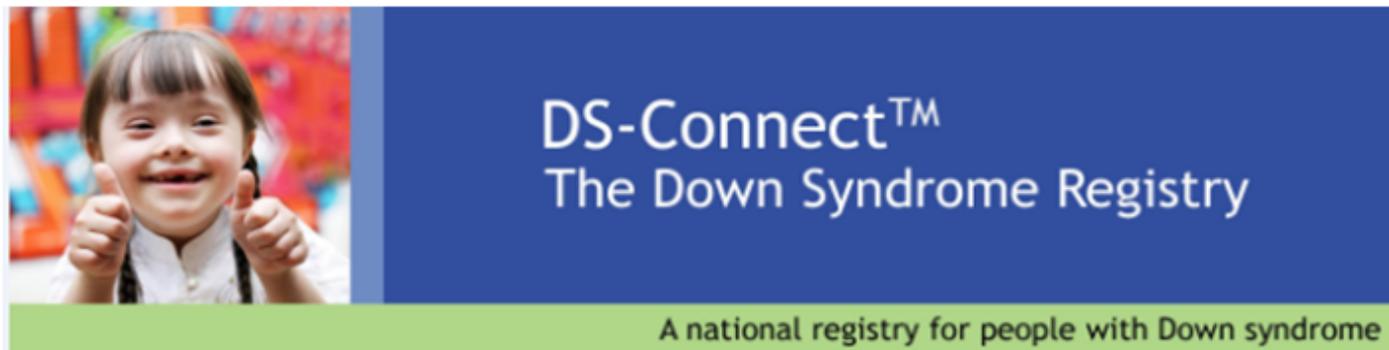
Her health information matters.

DS-Connect™: The Down Syndrome Registry
Get DS-Connected: dsconnect.nih.gov

Eunice Kennedy Shriver National Institute of Child Health and Human Development



Publicity: DS-Connections Quarterly Newsletters



DS-Connections: Quarterly Update | February 2014

Dear XXX (First name),

Thank you for registering at [DS-Connect™: The Down Syndrome Registry!](#)

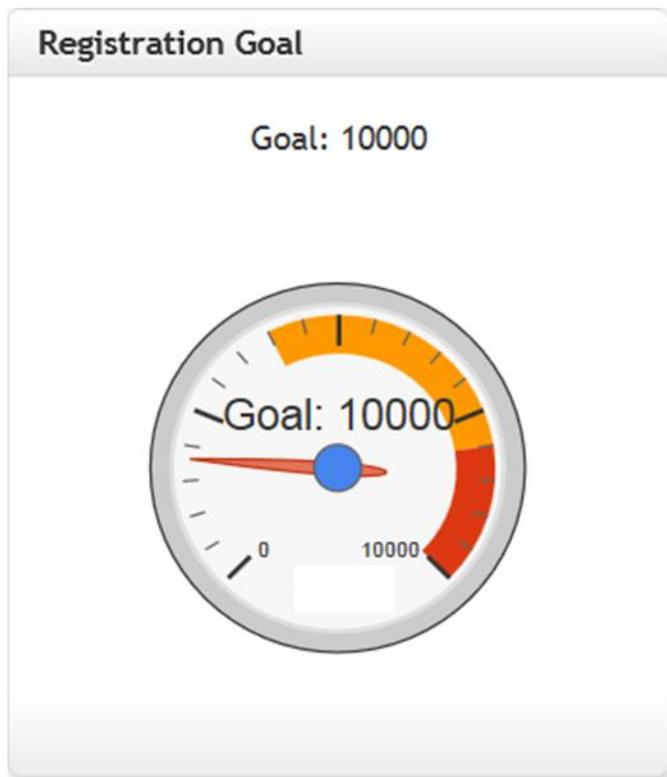
About 1,350 families have registered with DS-Connect™ since September 6, 2013. Check out the U.S. map below for details. You can also view this map in real time. Just [login to your account](#) and select "Registry Data."



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

- ❖ Ongoing goals:
 - Involve more families, clinicians, and scientists in Down syndrome research
 - Use research to inform parents and families about latest research-supported facts





The Goal: Helping all individuals reach their full potential.

<http://downsyndrome.nih.gov>

<https://DSConnect.nih.gov>



Why should I sign up?

- ❖ **All** DS related health information matters
- ❖ You can find healthcare providers
- ❖ You can update your information at any time
- ❖ You can print out your survey answers and share them with medical providers
- ❖ You can help us update DS growth charts
- ❖ You can let NIH and scientists know about health needs of individuals with DS of all ages
- ❖ You can connect with researchers for future clinical studies

- ❖ **It only takes a few minutes...**

DS-Connect™

The Down Syndrome
Registry

<https://DSConnect.nih.gov>



National Institutes of Health



Get DS Connected!

Advance science,
improve lives.



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



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

Share her health information—
help others.



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



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

<https://DSConnect.nih.gov>



Acknowledgments

- ❖ External Scientific Reviewers
- ❖ Registry Governance Board
- ❖ Registry Operations Board
- ❖ Down Syndrome Consortium
- ❖ Families and individuals with DS
- ❖ NIH Down Syndrome Working Group
- ❖ DS-Connect™ development team:

Consortium Members

NIH National Institutes of Health
Down Syndrome Working Group

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

Jérôme Lejeune
FOUNDATION
research, care, advocacy

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

National Cancer Institute (NCI)

National Heart, Lung and Blood Institute (NHLBI)

National Institute of Mental Health (NIMH)

National Institute of Neurological Disorders and Stroke (NINDS)

National Institute on Aging (NIA)

aaidd

AUCD
ASSOCIATION OF UNIVERSITY CENTERS ON DISABILITIES

DOWN SYNDROME AFFILIATES IN ACTION

DMIG-USA
Down Syndrome Medical Interest Group-USA

Lumind
Down Syndrome Cognition Research FOUNDATION

GLOBAL DOWN SYNDROME FOUNDATION

International Mosaic Down Syndrome Association

NATIONAL DOWN SYNDROME CONGRESS

national down syndrome society
ndss

Research Down Syndrome

Special Olympics

- Sujata Bardhan, PhD
- Aubrey Callwood
- Debbie Jae, MS
- Lisa Kaeser, JD
- Vanessa Rangel Miller, MA, MBA
- Christina Stile
- Yvonne Maddox, PhD



NIH Research Plan on Down syndrome

- ❖ Original NIH Research Plan released in 2007
- ❖ 2014 revision: update the goals and objectives for NIH-supported research on Down syndrome
- ❖ Led by NIH DS Working Group with input from the scientific, family, and advocacy communities
- ❖ 5 major areas: Pathophysiology of DS and Disease Progression; Diagnosis, Screening, and Functional Measures; Treatment and Management; Down Syndrome and Aging; and Research Infrastructure
- ❖ <http://www.nichd.nih.gov/news/releases/Pages/030414-DS-research-plan.aspx>
- ❖ Stay tuned for the final draft!