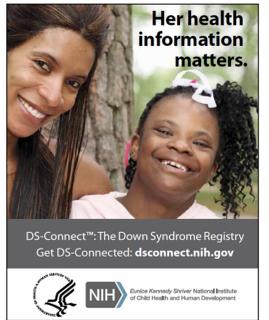
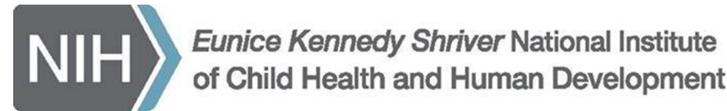


# Down Syndrome Resources from the National Institutes of Health: Research Projects Supported by the DS-Connect® Registry



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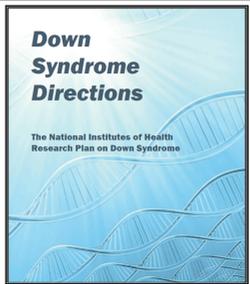


## BACKGROUND

**Background:** The National Institutes of Health (NIH) supports Down syndrome (DS) research to better understand the condition and develop treatments and interventions. NICHD/NIH launched *DS-Connect®: The Down Syndrome Registry* (<http://DSConnect.nih.gov>) in 2013 to facilitate information sharing among persons with DS, families, and researchers. It consists of a series of online surveys to collect demographic information as well as health information about people with DS. The Registry has over 3700 participants, and has been supported by the Down Syndrome Consortium, composed of self-advocates, family members, DS organizations and foundations, and the NIH. In 2014, the professional portal was launched to allow investigators to access the de-identified data for research purposes. The registry also enables researchers to use de-identified data to develop studies on the etiology, natural history, and treatments for DS and associated conditions. In addition, NIH has published updated research objectives for DS in *Down Syndrome Directions: The NIH Research Plan on Down Syndrome*.

## NIH Research Plan on DS

- 2014 revision includes 5 major sections:
  - Pathophysiology of DS and Disease Progression
  - Down Syndrome-Related Conditions: Diagnosis, Screening, and Functional Measures
  - Treatment and Management
  - Down Syndrome and Aging (new)**
  - Research Infrastructure
- Posted at: [https://www.nichd.nih.gov/publications/pages/pubs\\_details.aspx?pubs\\_id=5865](https://www.nichd.nih.gov/publications/pages/pubs_details.aspx?pubs_id=5865)



## DS-Connect®



**DS-Connect®: The Down Syndrome Registry**  
 A secure, confidential, online survey tool to collect basic health information about people with Down syndrome. Now available on a responsive web template, accessible from a mobile device.  
**Goals of DS-Connect®:**

- To identify the various phenotypic manifestations of 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

## DS-Connect® Professional Portal

Link to set-up a Professional Account from the Home page

Registration Goal: Goal 10000, Current: 3716

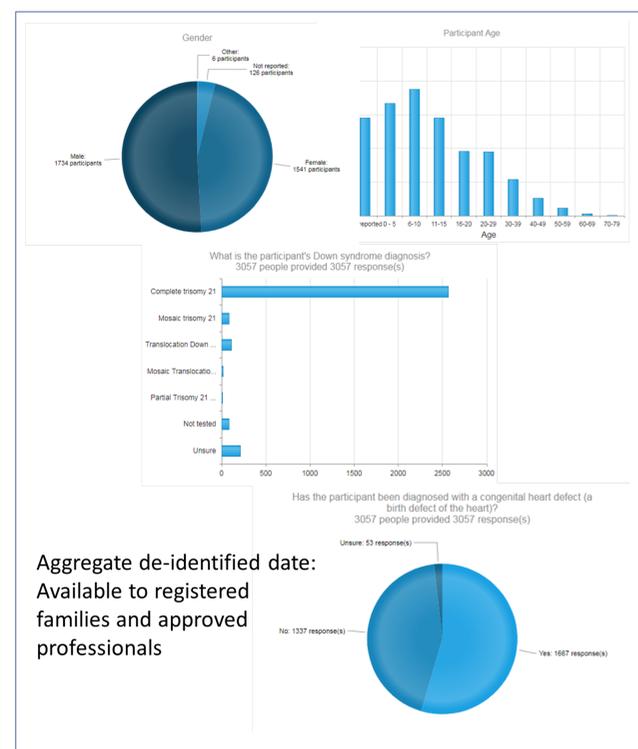
Over 270 individuals have signed up for a professional account

Registered Professionals

## Professional Portal: Tiers of Access

- Access Level 1:** View data identical to Registrants' view and perform basic searches of de-identified data
  - Access Level 2\*:** More customized searches, statistical analysis, publication or presentation.
  - Access Level 3\*:** For study recruitment, posting a study announcement, to propose a new survey module, or for commercial purposes.
- \*requires submission of proposal, purpose, and IRB status, and review by Research Review Committee for approval
- In the past 3 years:
    - 6 studies have been submitted for level 2 access
    - 21 studies have been submitted for level 3 access.
  - 18/21 level 3 studies have been approved by the RRC (86% success rate).
  - More than 7000 notifications have been sent to invite registrants to participate in research projects.**

## DS-Connect®: Explore the Data



## Supported Research Projects

- Published and Completed Survey-based studies:**
- Online survey of parent experiences in feeding their children with DS
  - Phone interview of parents to learn how children with DS find their way (navigate)
- Others:
- Studying sleep as a marker for brain development and learning in children with DS
  - Study of obesity and risk of diabetes in children with DS
  - Studies of early thyroid screening for infants to determine optimal screening schedule
  - Study of motor function and balance control in children with DS
  - Helping to recruit for University of Alabama Down syndrome Registry
  - Survey of opinions on medical research in DS
  - Study of gynecologic issues in women with DS
  - Survey of social and communication skills in children with DS
  - Recruitment for a virtual clinic: Down Syndrome Clinic to You
  - Recruitment for the ABC-DS study to identify risk factors for development of Alzheimer's Disease in adults with DS
- NEW: Online survey of readiness for transition to adult care for adolescents with DS and their families**

## "ABC-DS": Alzheimer's Biomarker Consortium-DS

**Goal:** Identify risk factors for Alzheimer's disease in adults with Down syndrome using markers (cognitive tests, brain MRI scans, genetics, blood tests)

- > 500 Down syndrome participants
- 2 awards made Sept 2015: \$37 M over 5 yrs:
  - Benjamin Handen (Univ of Pittsburgh); University of Wisconsin-Madison; Cambridge University, England; Alzheimer's Disease Cooperative Study.
  - Nicole Schupf (Columbia University); New York State Institute for Basic Research; Univ of California, Irvine; Kennedy Krieger Institute/JHU; Massachusetts General Hospital; Univ North Texas.

<https://www.nia.nih.gov/alzheimers/alzheimers-biomarkers-consortium-down-syndrome-abc-ds>

## New Congressional Directive for DS

- In the FY 2018 appropriations legislation, Congress directed NIH to develop a new trans-NIH initiative – involving, at a minimum, NICHD, NIA, and NCI – to study trisomy 21, with the aim of yielding scientific discoveries to improve the health and neurodevelopment of individuals with Down syndrome and typical individuals at risk for Alzheimer's disease, cancer, cardiovascular disease, immune system dysregulation, and autism, among others.
- NIH is working on a plan to implement this new mandate.

## Down Syndrome Consortium

A public-private partnership to foster communication and idea-sharing among the NIH, individuals with Down syndrome and their families, national organizations and foundations interested in DS: