



Government Funding for Down Syndrome Research Triples – What This Could Mean for Children & Adults with Down Syndrome and How You Can Help

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Eunice Kennedy Shriver National Institute
of Child Health and Human Development





What if researchers studied...

- ...why some babies with Down syndrome get leukemia?
- ...the ways children with Down syndrome communicate, learn and develop?
- ...the best strategies for kids with Down syndrome to get the right eyeglass prescription?
- ...treatments for skin disorders in adults with Down syndrome?
- ...how best to measure sleep apnea in a home setting rather than going to a hospital or clinic?

They are! These and other studies are being funded through...



NEWS RELEASES

Wednesday, September 25, 2019

New NIH grant awards boost funding for research on Down syndrome

INCLUDE project aims to address conditions of Down syndrome and apply knowledge to larger population.



The National Institutes of Health has awarded approximately \$35 million in new grants in fiscal year (FY) 2019 to advance research on Down syndrome through the [Investigation of Co-occurring Conditions Across the Lifespan to Understand Down Syndrome](#) (INCLUDE) project. These awards bolster total funding for Down syndrome research in FY 2019 to an estimated \$77 million. INCLUDE is a trans-NIH effort with participation from many NIH institutes and centers. Research funded by INCLUDE will investigate critical health and quality-of-life needs for individuals with Down syndrome, while at the same time explore co-occurring conditions with individuals who do not have Down syndrome.

NIH awards ~ \$77 M in funding for Down syndrome research this year!

<https://www.nih.gov/news-events/news-releases/new-nih-grant-awards-boost-funding-research-down-syndrome>



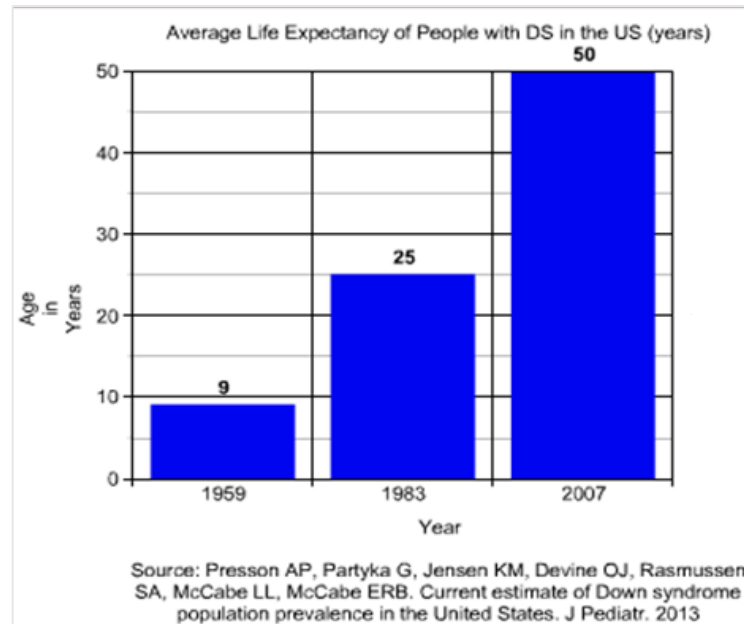
Outline

- National Institutes of Health (NIH)
- The INCLUDE Research Initiative
 - History
 - Research being funded this year
- NIH resources for Down syndrome
 - Down Syndrome Research Plan
 - DS-Connect[®]: The Down Syndrome Registry



Why is research important?

- People with Down syndrome are living longer, largely due to improved medical treatments



- In 1959, the life expectancy of a person with DS in the US was 9 years.
- In 1983, the life expectancy of a person with DS was 25 years.
- In 2007, the life expectancy of a person with DS was 50+ years—
an increase of >450% from 1959!

What is the National Institutes of Health (NIH)?

- The U.S.'s national medical research agency
- The world's largest funder of biomedical research
- Made up of 27 separate research institutes and centers
- Director since 2009: Dr. Francis Collins
- 85% of funds go to university research institutions based on peer review of grant applications



NIH leads the 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)

National Institute of Mental Health (NIMH)

National Institute of Neurological Disorders and Stroke (NINDS)

National Institute on Aging (NIA)

National Institute on Minority Health and Health Disparities (NIMHD)

National Institute of Dental and Craniofacial Research (NIDCR)

National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)

National Institute on Deafness and Other Communication Disorders (NIDCD)

National Human Genome Research Institute (NHGRI)

National Institute of Allergy and Infectious Diseases (NIAID)

National Center for Advancing Translational Sciences (NCATS)

Self-advocates



**DOWN SYNDROME
AFFILIATES IN ACTION**



alzheimer's  association®





New Congressional Directive for DS

- In the 2018 budget legislation for NIH:
 - Develop a new **trans-NIH** initiative 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
 - Leukemia
 - Heart defects
 - Immune system dysregulation
 - Autism
 - and other conditions
- Congress provided ~\$23 in additional research money in 2018 for a new trans-NIH research initiative

Protected from:

- Many cancers
- Heart disease and heart attacks



INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrom**E**)

3 components:

- 1. Conduct targeted, high-risk, high-reward basic science studies on chromosome 21.**
- 2. Assemble a large study population of individuals with Down syndrome across the lifespan.**
- 3. Include individuals with Down syndrome in existing and future clinical trials.**



2018 INCLUDE Funding for DS

- **~\$23 M supported 49 supplements to existing projects**
- All 3 components addressed
- **General areas:**
 - Leukemia and immune system
 - Language
 - Development
 - Pulmonary function
 - Sleep
 - Genomics
 - Aging and Dementia



2019 INCLUDE Funding for DS

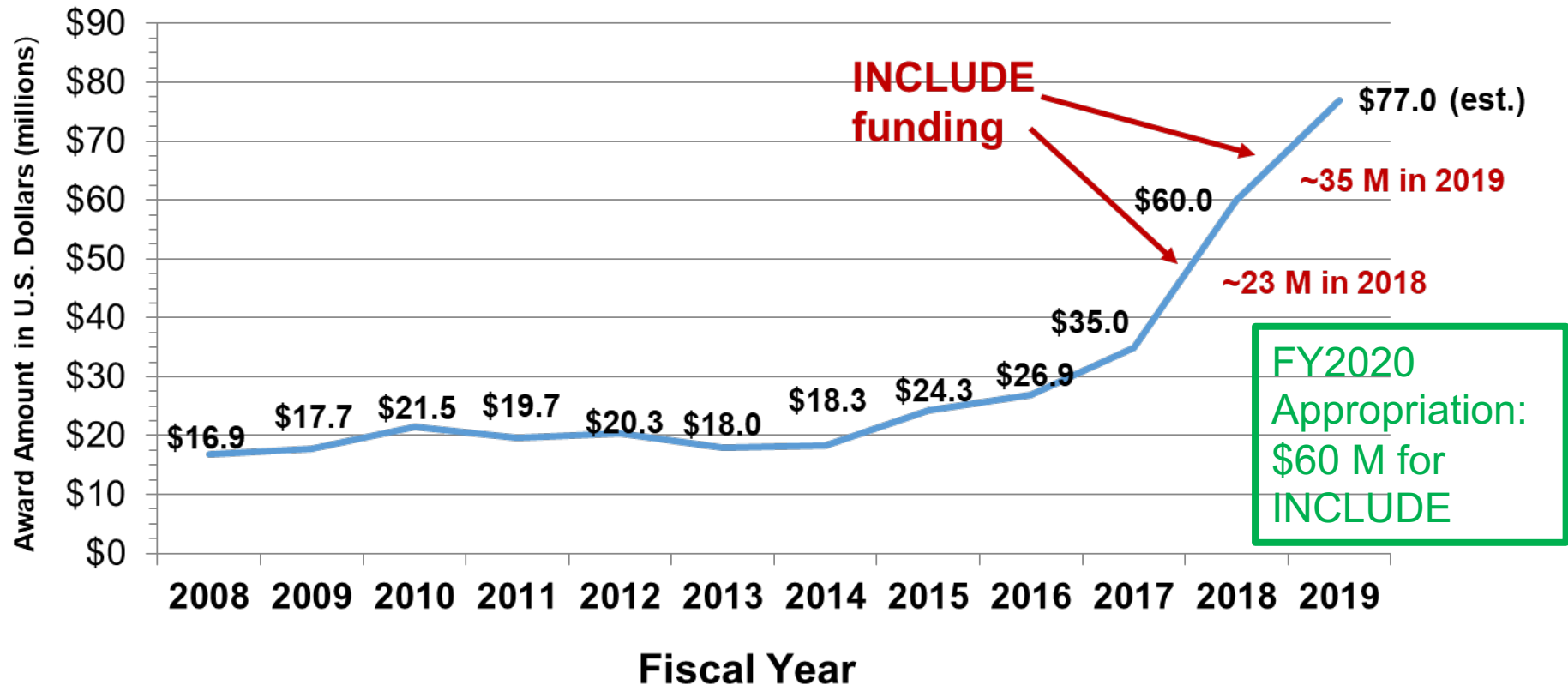
- **~\$35 M supporting 43 projects**
- **Basic science:**
 - Silencing genes on chromosome 21 to understand brain development
 - Studies to understand why $\sim\frac{1}{2}$ of infants have a heart defect
- **Cohort development:**
 - Cognitive and developmental tests for future clinical trials
 - Effects of chemotherapy used to treat leukemia on the heart
 - Hearing in a noisy environment
 - Strategies to recruit underrepresented minorities into research
- **Clinical trials:**
 - Medications for ADHD in children with attention problems
 - Medications to treat sleep apnea in children
 - A medication for Alzheimer's disease in adults
- **Training:** support for up to 18 trainees and junior researchers

URL: <https://www.nih.gov/include-project>



Total NIH Funding for Research on Down Syndrome

2008 – 2019

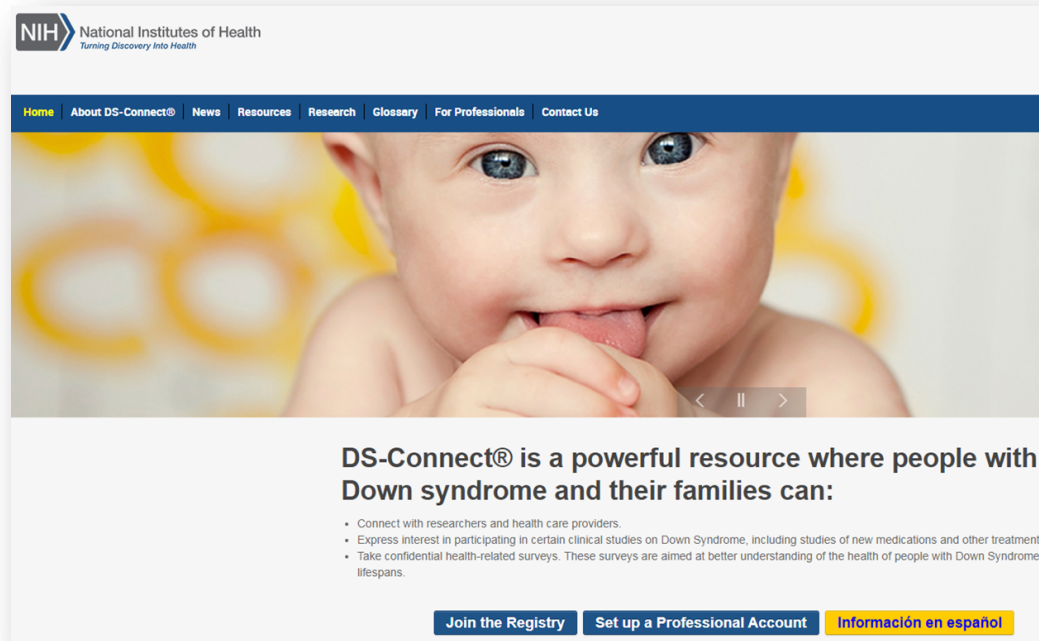




Building a large cohort for natural history and biomarker studies

- Several projects: Genomic predictors of heart defects, leukemia, and other features in individuals with Down syndrome
- Use DS-Connect[®]: The Down Syndrome Registry for recruitment

<http://DSConnect.nih.gov>



Alzheimer's Disease in DS

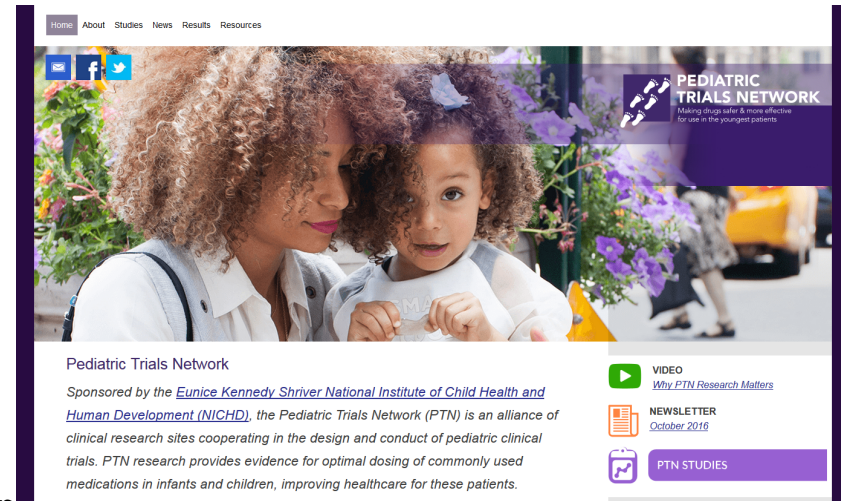


- Many people with DS begin to show symptoms of Alzheimer's disease (AD) in their 50s/60s
- Alzheimer's Biomarkers Consortium of Down Syndrome (ABC-DS):
 - Funded by NIH (NIA and NICHD): ~\$46 M over 5 years
 - > 500 volunteers who have Down syndrome, plus healthy siblings
 - Age >25 years
 - Studies of biomarkers that may help diagnose Alzheimer's—even before symptoms appear:
 - Cognitive tests
 - Brain scans (amyloid, tau)
 - Genetic studies
 - Blood biomarkers
 - <https://www.nia.nih.gov/alzheimers/alzheimers-biomarkers-consortium-down-syndrome-abc-ds>
- INCLUDE projects to understand lifestyle factors and lay the groundwork for clinical trials of AD in DS
- New research in 2019:
 - Clinical trial to test preventive treatments for dementia in adults with DS



NICHD's Pediatric Trials Network (PTN)

- Studies the formulation, dosing, efficacy, and safety of drugs and medical devices used in children
- Conducts clinical trials for many different conditions
- Supported by NICHD through the Best Pharmaceuticals for Children Act (BPCA), legislation that mandates drug studies in children to enable FDA labeling
- PTN clinical research sites are located at over 100 universities and children's hospitals across the U.S.
- **The PTN will partner with INCLUDE to support drug studies in individuals with Down syndrome**
- **Additional goal is to train investigators to conduct research involving people with DS**



See www.Pediatrictrials.org

Health Information

Grants & Funding

News & Events

Research & Training

Institutes at NIH

About NIH

[Home](#) » [Research & Training](#) » [Medical Research Initiatives](#)

THE INCLUDE PROJECT

Coming up in 2020

INCLUDE Project Research Plan



- More funding opportunities!
- Clinical trials infrastructure/training for investigators
- Workshops:
 - May 2020: Clinical Trials in DS
 - Nov. 2020: Opportunities in Basic Science in DS
- **Give us feedback!**

URL: <https://www.nih.gov/include-project>



New Funding Opportunity Announcements (FOAs)

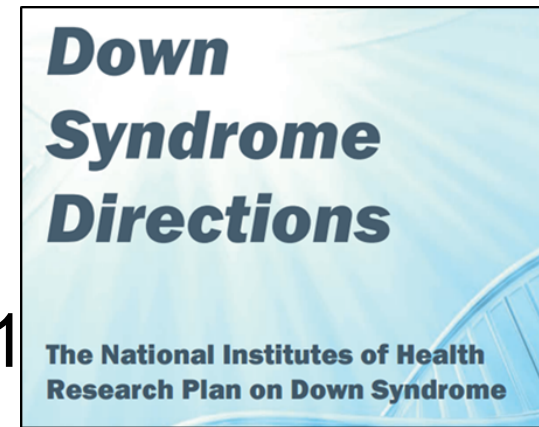
- The NIH has re-issued 3 Request for Applications (RFAs) in 2020:
 - 1. Transformative Research Award (Transformative R01)
 - 2. Clinical Trial Readiness Award (R21)
 - 3. Clinical Trials for Co-Occurring Conditions in Down Syndrome (R61/R33)
- **2 New RFAs:**
 - 4. Data Coordinating Center (U2C)
 - 5. Secondary Data Analysis (R03)
- **Other New** Notices of Special Interest (NOSIs):
 - 6. Mentored Fellowships and Career Development Awards
 - 7. Model organism development
 - 8. New grant applications (R01) and competitive supplements (R01) for DS research
 - 9. Administrative supplements to existing grants

For details:
<https://www.nih.gov/include-project/funding>



NIH Research Plan on Down Syndrome: Down Syndrome Directions

- Goal: Develop research goals and objectives for NIH research
- Plan to update the 2014 plan for 2021
- 5 major Research areas:
 - Pathophysiology of Down Syndrome and Disease Progression
 - Screening, Diagnosis and Functional Measures
 - Treatment and Management
 - Research Infrastructure
 - Down Syndrome and Aging



Look for calls to provide public input through Requests for Information!

How you can help...



Sign up for DS-Connect® (<http://DSConnect.nih.gov>)

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

The screenshot shows the DS-Connect website homepage. At the top, there's a navigation bar with links for LOGIN, JOIN, and NEED HELP?, along with flags for English and Spanish. Below this is the NIH logo and the text 'National Institutes of Health Turning Discovery Into Health'. A secondary navigation bar includes links for Home, About DS-Connect®, News, Resources, Research, Glossary, For Professionals, and Contact Us. The main content area features a large video player showing a smiling man. Below the video, the text reads: 'DS-Connect® is a powerful resource where people with Down syndrome and their families can:'. This is followed by a bulleted list of benefits: connecting with researchers and health care providers, expressing interest in clinical studies, and taking confidential health-related surveys. At the bottom, there are three buttons: 'Join the Registry', 'Set up a Professional Account', and 'Información en español'.

NIH National Institutes of Health
Turning Discovery Into Health

LOGIN JOIN NEED HELP?  

Forgot login?
Unblock Account

Home | About DS-Connect® | News | Resources | Research | Glossary | For Professionals | Contact Us

DS-Connect® is a powerful resource where people with Down syndrome and their families can:

- Connect with researchers and health care providers.
- Express interest in participating in certain clinical studies on Down Syndrome, including studies of new medications and other treatments.
- Take confidential health-related surveys. These surveys are aimed at better understanding of the health of people with Down Syndrome across their lifespans.

Join the Registry Set up a Professional Account Información en español

Launched Sept. 6, 2013



A partnership with families...

➤ Access the health care provider list



[Questionnaire](#) [Healthcare Providers](#) [Growth Measurements](#) [Account Info](#) [Attachments](#) [Contacts](#) [Newsletters](#)

[Notes](#)

Search the directory for your health care provider(s) by name, facility name, city or state (2 letter abbreviation). If your health care provider is not in the directory, use the Add a New Health Care Provider link to add them in the directory. Note that you will need to search for each physician individually before the Save button will be enabled.

Please note that this list of healthcare providers does not imply endorsement or recommendation of their services.

Name/Institution

Specialty

City


State

Country



Search

Leave blank to list all

[Add a New Health Care Provider](#)

Click the  to add the healthcare provider to your account.

~764 now Listed!

Name	Specialty	Sees Adults or Youth?	Institution	City	State	Country	
John Avallone	Ophthalmology		Ophthalmology Associates	Arnold	MD	UNITED STATES	 My Health Care Provider
Mihee Bay	Developmental Pediatrics		Kennedy Krieger Institute	Baltimore	MD	UNITED STATES	 My Health Care Provider



A partnership with families...



➤ Print out the participant's medical history

Questionnaire	Healthcare Providers	Growth Measurements	Account Info	Attachments	Contacts
Notes					



Initial Health Questionnaire



Completed: 2013-09-25

PRINT



Sleep Questionnaire

PRINT

Jane Smith

DOB: 2011-01-01
Gender: Female
Registered By: Melissa Parisi
Relationship: Biological Parent

Initial Health Questionnaire

Diagnosis

- What is the participant's Down syndrome diagnosis?
Complete trisomy 21
- How was the diagnosis of Down syndrome made? (Select all that apply.)
Genetic testing in baby after birth (such as chromosome analysis, cytogenomic array, or fluorescence in situ hybridization (FISH))
- What was the participant's age in years when the diagnosis of Down syndrome was made?
At birth



A partnership with families...

- Access the healthcare recommendations
 - Personalized for the age of the person with DS



Health Care Guidelines For Individuals With Down Syndrome

[Children with Down Syndrome: Health Care Information for Families \(AAP\)](#)

[Age-Specific Health Care Checklist \(AAP\) \(144 KB PDF\)](#)

[Aging and Down Syndrome: A Health & Well-Being Guidebook \(NDSS\) \(8 MB PDF\)](#)

Links to AAP checklists

Health Care Information for Families of Children with Down Syndrome

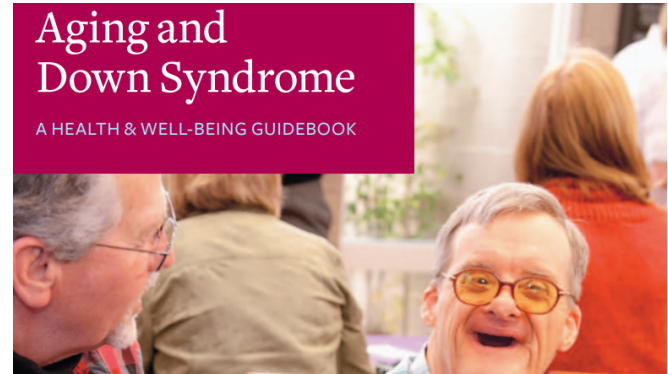
Child's Age: 13 to 21 Years or Older

- ☐ **Regular well-care visits (check-ups)**
It is important to have yearly well-care check-ups. These visits will assist in checking your child's health, giving shots, and answering questions about your child's health.
- ☐ **Monitor growth**
It is important to check growth at every visit. Measurements include height, weight, and body mass index (BMI). These measurements are very important to assessing the overall health of the child. Discuss diet, activity level, and growth. Your child's doctor can help with question about any need for vitamins or supplements.
- ☐ **Immunizations (shots)**

Links to NDSS guidebook

Aging and Down Syndrome

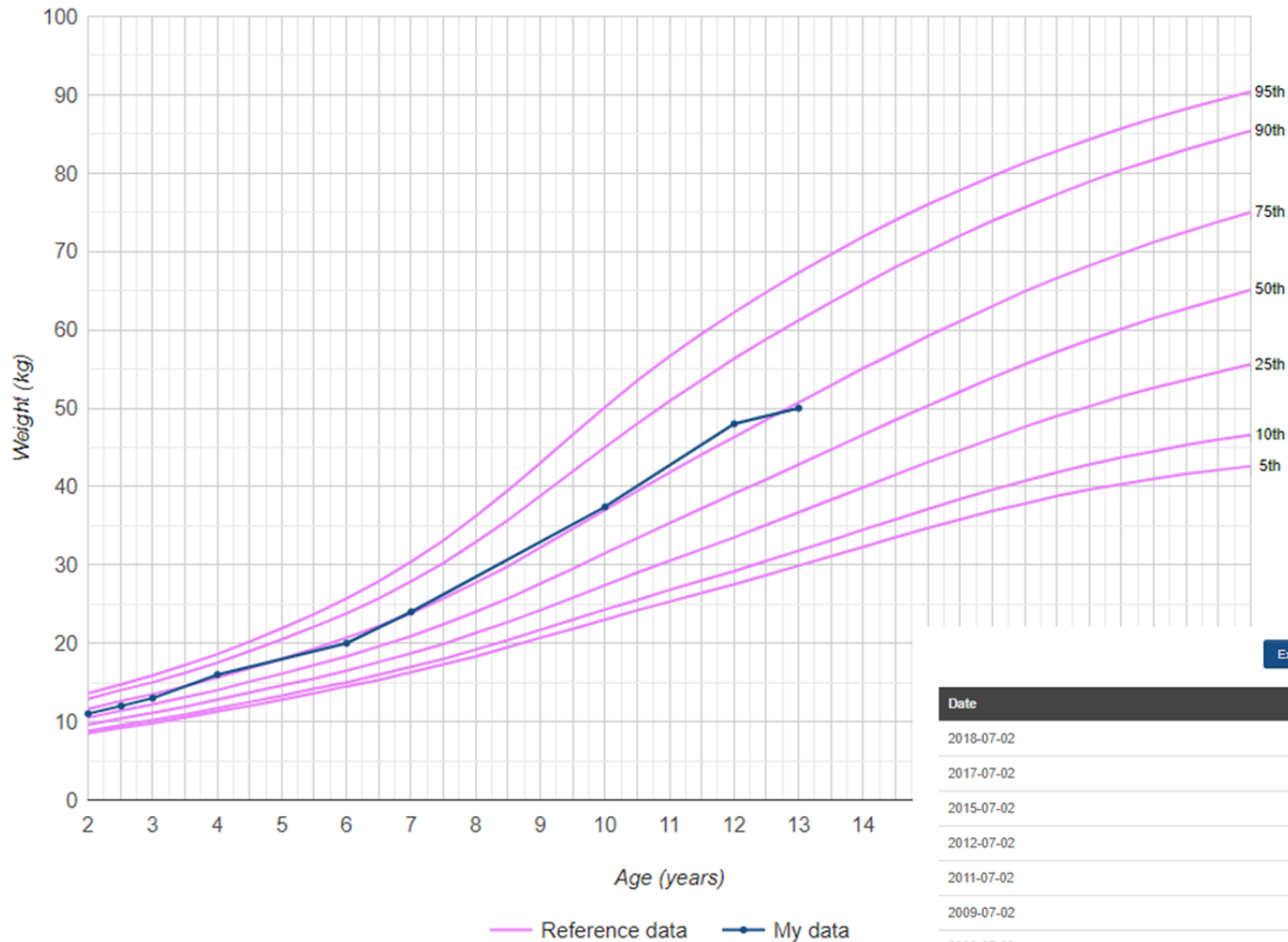
A HEALTH & WELL-BEING GUIDEBOOK





Plot and print your child's growth charts and data

Growth Charts for Children with Down Syndrome 2 to 20 years: Girls Weight-for-age percentiles



Export as PDF

Export as Image

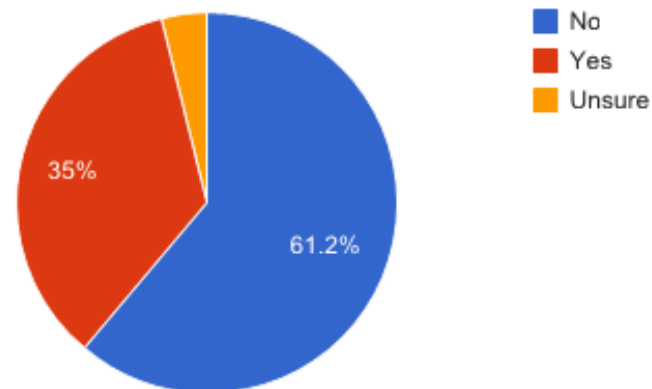
Date	Age (years)	Weight (kg)
2018-07-02	13	50.00
2017-07-02	12	48.00
2015-07-02	10	37.40
2012-07-02	7	24.00
2011-07-02	6	20.00
2009-07-02	4	16.00
2008-07-02	3	13.00
2008-01-06	3	12.00
2007-07-02	2	11.00



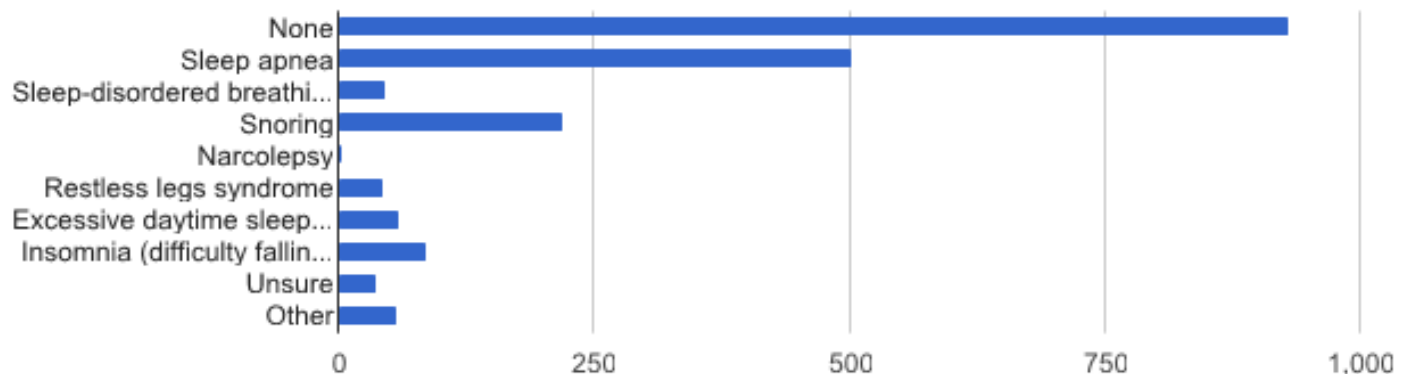
A partnership with families: Explore the data

Has the participant ever been diagnosed with any sleep problems? (1803 responses)

Example:
Sleep



Which of the following sleep problems have been diagnosed? (Select all that apply.)



1649 people provided 1993 response(s)



Recently Launched: Medication Tracker

Medication and Supplement Tracker

[Add](#)

[List](#)

Select "Add" to add a new prescription or over-the-counter medicine, vitamin, or herbal supplement. Select "Edit" if you stopped taking or need to change information about an item that is already in the list.

Current Medications and Supplements

Add Medication or Supplement

Sujata Bardhan

To help inform research, DS-Connect is collecting information about medication and supplement use in people with Down syndrome.

Type the name of a medication, vitamin, or herbal supplement in the search box and select "Search" to find the item you want to add to your list.

What are you taking? *

Search

tylenol

☐ Tylenol


☐ Acetaminophen

☐ Other



Portal for Professionals: Recruit for studies

[Home](#) [About DS-Connect®](#) [News](#) [Resources](#) [Research](#) [Glossary](#) [For Professionals](#)

A photograph of a woman with blonde hair smiling next to a young boy with blonde hair and Down syndrome. The boy is holding a small object in his hands. They are in a room with a window and some decorations.

[How do I turn on subtitles/captions and specify their language?](#)

[Join the Registry](#) [Set up a Professional Account](#)

DS-Connect® is a powerful resource where people with Down syndrome and their families can:

- Connect with researchers and health care providers.
- Express interest in participating in certain clinical studies on Down Syndrome, including studies of new medications and other treatments.
- Take confidential health-related surveys. These surveys are aimed at better understanding of the health of people with Down Syndrome across their lifespans.

- Professionals can apply to use DS-Connect to recruit for their studies
- If approved, they have 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



What does it mean to be in a research study with DS-Connect®?

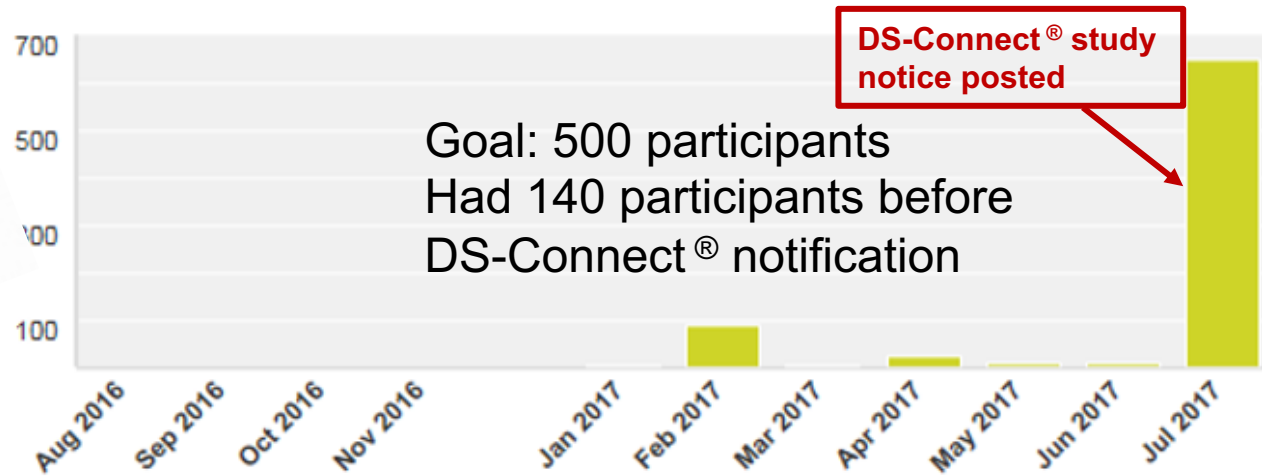
- ☐ Take a survey of your experiences
- ☐ Answer a questionnaire about health issues
- ☐ Record data from an activity tracker (e.g., Fitbit)
- ☐ Participate in a clinical study of a intervention
- ☐ Be in a clinical trial for a new drug or medication
- ☐ Participate in an “INCLUDE” study
- ☒ **Any of the above! It's your choice**





Recruitment Assistance from DS-Connect[®]

Dr. Amy Lewanda at Children's National studying use of nutritional supplements in children with DS



THE JOURNAL OF PEDIATRICS • www.jpeds.com

ORIGINAL
ARTICLES

Patterns of Dietary Supplement Use in Children with Down Syndrome

Amy Feldman Lewanda, MD, Margo Faust Gallegos, CGC, and Marshall Summar, MD

Conclusions Almost one-half of parents surveyed administer or have administered supplement(s) to their children with Down syndrome. Many of the supplements have concerning ingredient profiles and are given to children too young to articulate potential ill effects. Providers need to be aware of these products and question families about their use. (*J Pediatr* 2018;201:100-5).



Spread the word about DS-Connect

- **Sign up!**
- Electronic Toolkit available:
 - Video Testimonial-YouTube
 - Flyers
 - Facebook info-cards
 - Badges and Tweets
- Help us reach our goal:
 - 10,000 by Dec 31, 2020

Registration Goal

Goal: 10000
Current: 4922



**Advance science,
improve lives.**

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

Eunice Kennedy Shriver National Institute
of Child Health and Human Development

<http://DSConnect.nih.gov>



What is missing?





Q & A

- **DS-Connect:** <http://DSConnect.nih.gov>
- **INCLUDE:** <https://www.nih.gov/include-project>
- Contact us! DSConnect@nih.gov
or parisima@mail.nih.gov



Melissa Parisi



Sujata Bardhan



Lisa Kaeser

On behalf of the entire NIH INCLUDE Team