The NIH INCLUDE Project

An Opportunity to Promote Health for All Individuals with Down Syndrome

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Global Research & Medical Care Roundtable
Down Syndrome Diagnosis Network
Outline

- The National Institutes of Health
- The INCLUDE Project
- DS-Connect® Registry
- Community Engagement and Outreach
Acknowledgements

Linda Garcia

• The NIH-wide Down Syndrome INCLUDE Working Group

• Investigators and Trainees

• Children and adults with Down syndrome and their families
The NIH

• The U.S.’s national medical research agency based in Bethesda, Maryland

• The world’s largest funder of biomedical research

• Made up of 27 separate research institutes and centers

• 90% of funds go to university research institutions based on peer review of grant applications
The INCLUDE PROJECT
INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome
The INCLUDE Project Goals

• Launched in 2018 under a Congressional Directive to address critical health and quality of life needs for those with DS.

• INCLUDE is investigating conditions that affect individuals with DS and the general population, such as Alzheimer’s disease, autism, cataracts, celiac disease, congenital heart disease, and diabetes.

• The project will also increase the number of investigators/trainees studying DS.

• The project will engage with those with DS and their families from diverse backgrounds.
INCLUDE Project

Three components:

1. Conduct targeted, high-risk, high-reward basic science studies on chromosome 21.

2. Build a large cohort of individuals with Down syndrome for comprehensive analysis and biomarker evaluation.

3. Include individuals with Down syndrome in existing and future clinical trials.
The NIH has invested $258 million in the past 5 years on 270 new projects for INCLUDE.
What has INCLUDE done so far?

- Study the causes of acute lymphoblastic leukemia in children with DS
- Study hearing loss across the lifespan in those with DS
- Study language acquisition and articulation in children with DS
- Develop and validate cognitive measures in the NIH Toolbox for children with DS.
- Develop a Behavior Inventory for Down Syndrome (BIDS) for children and adolescents with DS in English and Spanish.
- Study the causes of congenital heart disease in infants with DS, and whether heart surgery impacts their neurodevelopment and behavior
- Study a cohort of adults with DS to identify biomarkers of neurodegeneration and risk and resilience factors for Alzheimer's disease
NIH finalized the Research Plan for its INCLUDE Project and related DS research.

**Highlights of the plan include the following:**
- Public and DS community input in the new and revised goals and objectives
- Emphasis on increasing diversity of researchers and research participants
- New information about DS research training
- Detailed review of NIH research projects conducted between 2014 and 2020, and a bibliography with 590+ scientific articles
- Findings from research on COVID-19 in individuals with DS

Available for download on the INCLUDE Project website: nih.gov/include-project/include-project-down-syndrome-ds-research-plan
Improving the quality of life of people with Down syndrome

The INCLUDE Data Coordinating Center (DCC) is making it easier for scientists and the Down syndrome community to work together. Matching the latest technology with shareable resources, researchers use the Data Hub to enhance healthcare and change lives.

Learn More  Contact us
Thirteen Clinical Trials Funded by INCLUDE

Sleep & Apnea (OSA)
- Medications for OSA to improve cognition in children with DS
- Effects of hypoglossal nerve stimulation on cognition and language in DS
- Positive airway pressure for OSA in children with DS
- Home Sleep Apnea Testing Compared to In-lab Polysomnography for the Evaluation of OSA in Children with DS
- Self-Supporting Nasopharyngeal Airway Treating Upper Airway Obstruction in Hypotonia
- A Personalized Surgical Approach for the Treatment of Children with Obstructive Sleep Apnea and Small Tonsils
- Randomized Controlled Trial of Oxygen Therapy in Children and Adolescents with DS and OSA

Alzheimer's Disease & Aging
- Clinical trials to prevent Alzheimer's Disease in DS
- Addition of GM-CSF/sargramostim treatment to improve cognition in DS
- The Impact of Weight Loss on Alzheimer's Disease Risk in Adults with DS

Immune System Dysregulation
- JAK inhibition for treatment of DS skin conditions

Neurodevelopment
- Mechanistic investigation of therapies for Down Syndrome Regression Disorder
- Evaluating assessment and medication treatment of ADHD in children with DS
DS-Connect®: The Down Syndrome Registry

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

• Join DS-Connect and encourage other families to sign up!

Help us reach our goal:
• 10,000 participants by September 2023, the 10-year anniversary of the Registry

Launched 2013:
~5,868 registrants globally as of September 2023

dsconnect.nih.gov
Participants in the DS-Connect® Registry
How secure is it?

- Meets stringent data security requirements to protect personally identifiable information
  - ~250 security controls are checked regularly (FISMA moderate level)
  - Information is encrypted
  - Password requirements (at least 8 characters long, 1 upper case, 1 lower case, 1 symbol, 1 number)
    - Example: P@ssword3
  - Passwords must be changed every 10 months
  - No social security info is collected
  - No personal bank account info is collected
Multiple Survey Modules

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

✓ Basic Health Survey
✓ Sibling Survey
✓ Thyroid Survey
✓ Heart Survey
✓ Sleep Survey
✓ Skeletal Survey
✓ Gastrointestinal Survey
✓ Diabetes Survey
✓ Celiac Disease Survey
✓ Leukemia Survey
✓ Development Survey
✓ Prenatal and Birth Survey
✓ Adulthood Survey
✓ Men’s Health Survey
✓ Women’s Health Survey
✓ Transition to Adulthood Survey

Available to adults

Survey for 12-30 yo

dsconnect.nih.gov
What does it mean to participate in research?

- Take a survey of your experiences
- Answer a questionnaire about health issues
- Record data from an activity tracker or watch
- Participate in a clinical study of an 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
Resources on DS-Connect® Registry
Access the health care provider list

~864 now listed

dsconnect.nih.gov
Access the Health Care Recommendations personalized for the person with DS

Children with Down Syndrome: Health Care Information for Families (AAP)
Links to AAP checklists

Medical Care Guidelines for Adults with Down Syndrome

Aging and Down Syndrome: A Health & Well-being Guidebook (NDSS)

Alzheimer's Disease & Down Syndrome: A Practical Guidebook for Caregivers (NDSS)

dsconnect.nih.gov
Find Clinical Trials

Find Down Syndrome Clinical Trials

Search for NIH-funded clinical trials related to Down Syndrome

Filter by:

Location: 
Age: 

Search  Clear  See all NIH-funded Down Syndrome clinical trials

To search all clinical trials, visit clinicaltrials.gov

dsconnect.nih.gov
Community Engagement & Outreach

2023

2024

Investigator Workshop

Advancing Chicanos/Hispanics & Native Americans In Science

American College of Medical Genetics and Genomics

American Society for Human Genetics

Annual Biomedical Research Conference for Minority Students

SACNAS

ASHG

ABRCMS

Global

NDSC

DSDN

New York City Buddy Walk

Buddy Walk

DSDN

DSNDN

Global

NEW YORK CITY BUDDY WALK

Buddy Walk

Investigator Workshop

Advancing Chicanos/Hispanics & Native Americans In Science

American College of Medical Genetics and Genomics

American Society for Human Genetics

Annual Biomedical Research Conference for Minority Students

SACNAS

ASHG

ABRCMS

Global

NDSC

DSDN

New York City Buddy Walk

Buddy Walk
First INCLUDE Project DEIA Webinar held last month!

Visit the NIH INCLUDE Website to access the recording: nih.gov/include-project

Building trust and establishing positive relationships between researchers and the DS community
Learn more about DS-Connect®, The DS Registry

1. Scan the QR Code to complete the form
2. After submitting, expect to receive an email from our DS-Connect Team to help you join the registry!
Thank You!