Promising Research at the NIH

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Global Down Syndrome Research and Medical Care Roundtable
June 25, 2020
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…
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.
Outline

• National Institutes of Health (NIH)
  • History of the INCLUDE Research Initiative
  • Research being funded by INCLUDE

• What you can to help
  • Sign up for DS-Connect®: The Down Syndrome Registry
  • Give us feedback: Down Syndrome Research Plan
  • Volunteer to participate in research
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

Self-advocates
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 M in additional research money in 2018 for a new trans-NIH research initiative

Protected from:
- Many cancers
- Heart disease and heart attacks
What does it mean to be a “trans-NIH” initiative?

18 different institutes are involved in the INCLUDE Project, and they cover all the major co-occurring conditions in DS.
INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome)

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.
INCLUDE Funding for DS

- **Basic science:**
  - Silencing genes on chromosome 21 to understand brain development
  - Studies to understand why ~½ 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
  - Medications to treat autoimmune skin conditions

- **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

Award Amount in U.S. Dollars (millions)

Fiscal Year


Funding: $16.9, $17.7, $21.5, $19.7, $20.3, $18.0, $18.3, $24.3, $26.9, $35.0, $60.0, $86.0

INCLUDE Funding

FY2020 Appropriation: $60 M for INCLUDE

35 M in 2019
23 M in 2018
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
- 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
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
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.

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.

Launched Sept. 6, 2013
DS-Connect® Registrants: North America

Over 5000 registrants globally as of June 25, 2020
A partnership with families...

Access the health care provider list

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.

<table>
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<tr>
<th>Name</th>
<th>Specialty</th>
<th>Sees Adults or Youth?</th>
<th>Institution</th>
<th>City</th>
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<td>Baltimore</td>
<td>MD</td>
<td>UNITED STATES</td>
</tr>
</tbody>
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Click the + to add the healthcare provider to your account.

~764 now Listed!
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

http://DSConnect.nih.gov
Events in 2020

- More funding opportunities
- Clinical trials support and training for investigators
- A new data coordinating center for INCLUDE
- Workshops:
  - May 2020: Clinical Trials in DS
  - Nov. 2020: Opportunities in Basic Science and cohort development in DS

URL: https://www.nih.gov/include-project
New Funding Opportunity for COVID-19

• Notice of Special Interest (NOSI) regarding the Availability of Urgent Competitive Revisions and Administrative Supplements for Research on Coronavirus Disease 2019 (COVID-19) in Individuals with Down Syndrome for the INCLUDE Project

• **Notice Number: NOT-OD-20-129**

• **Goal:** to improve understanding and treatment of COVID-19 infection in those with DS given their increased risk of serious infection

• Supplements to existing grants

• Deadlines: July 13, Nov 12 (2020); Mar 12, July 12 (2021)

• Specific areas of focus for different NIH institutes

NIH Research Plans on Down Syndrome

- Goal: Develop research goals and objectives for NIH research
- We are updating the 2014 plan and combining it with the INCLUDE Research Plan for 2021
- Seeking comments on:
  - Effectiveness of the previous plans
  - Advances made since 2014
  - Remaining research gaps
  - Suggestions concerning new future research objectives
- Respond by July 10, 2020 to DownSyndrome@mail.nih.gov
What is missing?

We need you!

New knowledge and treatments

Research ideas

NIH, Foundations

Researchers

Research participants

We need you!
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

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On behalf of the entire NIH INCLUDE Team