ISSUE BRIEF: NIH Research on Down Syndrome

Legislative Request

Urge the House and Senate Appropriations Subcommittees on Labor, Health and Human Services to support full funding of the INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome (INCLUDE) project at the National Institutes of Health (NIH).

Background and Summary

Once largely neglected, the healthcare needs of those with Down syndrome are beginning to receive the attention they deserve. In just the last two decades, significant advances have been made, in part due to the support and leadership of the NIH in advancing biomedical and clinical research in the field of Down syndrome. Despite these advances, however, those with Down syndrome continue to face very significant unmet challenges across their lifespan, and public and private sector support for work in this area lags far behind that needed to improve their lives and reduce the burdens on those who care for them and society at large.

As part of the Consolidated Appropriations Act of 2018 (H.R. 1625), Congress directed the NIH Director to develop a new trans-NIH initiative to study trisomy 21, to yield scientific discoveries to improve the health and neurodevelopment of individuals with Down syndrome and typical individuals at risk for Alzheimer’s disease/dementia, autism, cataracts, celiac disease, congenital heart disease and diabetes. Applying the expertise and resources from multiple NIH Institutes and Centers, the INCLUDE project will conduct targeted, high-risk, high-reward basic science studies on chromosome 21; assemble a large study population of individuals with Down syndrome; and include individuals with Down syndrome in existing clinical trials.

The INCLUDE project was launched in June 2018 in support of the Congressional directive. On October 1, NIH announced it made 49 awards totaling $22.2 million in funding for INCLUDE research, bolstering total funding for Down syndrome research in FY2018 to an estimated $59 million. NIH anticipates that this level of support will be maintained through the fiscal year 2022. However, future funding for the INCLUDE project is dependent on Congress providing sufficient funding.

Fully funding the INCLUDE project will allow NIH to not only expand its current efforts on Down syndrome and commonly co-occurring conditions in individuals with Down syndrome that are also seen in the general population, but to build an integrated effort across NIH that will be truly transformative in these areas.

Key Messages

→ NIH should be commended for the work that has been done in recent years to expand our understanding of Down syndrome – NIH has focused on increased efforts on understanding
Alzheimer’s disease and other associated medical and psychiatric conditions in individuals with Down syndrome. In addition to the INCLUDE project, key initiatives in these areas include funding the “Biomarkers of Alzheimer’s Disease in Down Syndrome” initiative; working with the Down syndrome research community on the development of outcome measures for cognitive, behavioral, and physical measures for clinical trials; and coordinating with the NIH Down Syndrome Working Group to launch DS-Connect®, the Down syndrome patent registry.

→ Down syndrome is no longer a childhood condition, and effective research requires engagement and collaboration among all the NIH institutes and centers – At NIH, Down syndrome research is primarily been under the purview of the National Institute of Child Health and Human Development. Some individuals with Down syndrome are living to be nearly 80 years old, and there is much to learn from and about the medical and behavioral conditions that are associated with Down syndrome. The INCLUDE project will take advantage of the full range of existing resources across NIH, integrating the expertise of at least eighteen NIH institutes and centers.

→ The INCLUDE project will expand our knowledge not only of Down syndrome but of the commonly co-occurring conditions in individuals with Down syndrome that are also seen in the general population – For example, people with Down syndrome have an increased risk for certain medical conditions such as congenital heart defects, respiratory and hearing problems, Alzheimer's disease, childhood leukemia, and thyroid conditions. Many of them also have secondary neurobiological, behavioral and psychological conditions, such as Autism spectrum disorders. A Trans-NIH Initiative will provide new insights into many of the coexisting conditions in Down syndrome, such as Alzheimer’s disease, autism spectrum disorders, congenital heart disease, and how best to treat all people with those conditions, not just those with Down syndrome. The same holds for studying conditions that are rare in people with Down syndrome, such as solid tumors.

Answers to Questions about NIH Research on Down Syndrome

1. Why is Down syndrome best suited for a trans-NIH Initiative like the INCLUDE project?

   Down syndrome is no longer a childhood condition. Some individuals with Down syndrome are living to be nearly 80 years old, and there is much to learn from and about the medical and behavioral conditions that are associated with Down syndrome. Until now, the National Institute of Child Health and Development (NICHD) has been the lead agency on investigator-driven research awards in the area of Down syndrome. But the biology underlying the challenges that researchers and caregivers seek to improve is studied in multiple NIH Institutes, and structural barriers hamper support for these studies.

2. How much additional funding is needed to “fully fund” the INCLUDE project?

   The legislation that established the INCLUDE project stated that funding for the project would supplement, not supplant, existing NIH funding levels for Down syndrome research. There are no current estimates of how much it will cost to fund the project, although initial commitments called for providing $60 million for each year of the project above the fiscal year 2018 baseline funding. It should also be noted that Congress provided a $3 billion increase to the overall NIH
budget for the fiscal year 2018, as well as a $2 billion overall increase in the fiscal year 2019, of which part of that increased funding is being used to support the implementation of new initiatives like the INCLUDE project.

**Resources**

NIH INCLUDE Project – [https://www.nih.gov/include-project](https://www.nih.gov/include-project).

