



Down Syndrome Research Priorities

Overview

The extra copies of genes present on the 21st chromosome cause developmental problems and health issues in people with Down syndrome. However, due to advances in medical technology, individuals with Down syndrome are living longer than ever before. Today, as many as 80 percent of adults with Down syndrome reach the age of 60, and many live even longer. Biomedical research has advanced to the point where scientists now have a much better understanding of the genetic basis for cognitive and medical impairments associated with Down syndrome, and they have made unprecedented progress towards identifying effective treatments. Going forward, government funding and clinical research infrastructure support are vital to our efforts to translate research achievements into real treatments and therapies.

Co-Occurring Conditions

A lack of research on individuals with Down syndrome who have numerous co-occurring and/or simultaneous psychiatric and medical conditions has been an impediment to the development of clinical and behavioral treatments and interventions. According to the NIH, at least one-half of all children with Down syndrome also have one or more such conditions. During the early years of life and across their lifespan, these so-called “comorbid” conditions could have the potential to significantly affect cognitive function and overall health.

Down syndrome research could benefit significantly from the development of new tools and resources for measuring, for purposes of clinical outcomes, the differences in results related to co-occurring psychiatric or medical conditions and genetic differences among individuals. Authorizing NIH to develop classification and measurement tools for such conditions would significantly expedite ongoing efforts to develop effective treatments and interventions. Other psychiatric and medical conditions that could benefit from NIH’s partnership in the development of such tools include Alzheimer’s disease, childhood leukemia, congenital heart disease, autism spectrum disorders, sleep apnea and epilepsy, to name a few.

Alzheimer’s Connection

Scientists have discovered that the majority of individuals with Down syndrome develop the pathology of the disease while in the mid-40’s. They believe that studying Alzheimer’s in Down syndrome will provide new insights into how best to treat all people with Alzheimer’s disease, not just those with Down syndrome. In fact, the Department of Health and Human Service’s National Plan to Address Alzheimer’s Disease underscores the relationship between Down syndrome and Alzheimer’s disease by recognizing the need to improve access to long-term services and supports for individuals with Down syndrome. In addition, NIH recently published a new grant proposal to study biomarkers of Alzheimer’s disease in Down syndrome. The goal is

to identify the progression of Alzheimer's disease over the lifespan of adults with Down syndrome using clinical, cognitive, imaging, genetic, and biochemical indicators.

Completion of the biomarkers study could take up to five years and cost more than \$15 million, but NIH has only announced funding for the first year. Therefore, there is concern that future funding may not be sufficient to complete the study, or it could come at the expense of other high priority research investments in Down syndrome. This would be problematic, given that overall NIH funding for Down syndrome research-related projects, as well as investigator-driven research awards, has been stagnant in recent years.

It is important that NIH provide sufficient grant funding to complete the multi-year biomarkers study of Alzheimer's disease in individuals with Down syndrome, and to ensure that future funding does not come at the expense of ongoing and other priority research investments. In addition, effectively exploring the link between Down syndrome and Alzheimer's requires collaboration and resources across the various institutes of the NIH. 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. A more collaborative approach between NICHD, the National Institute of Aging (NIA) and the National Institute of Neurological Disorders and Stroke (NINDS) would facilitate biomedical advances in understanding both Down syndrome and Alzheimer's disease.

Public Private Partnerships

In 2006, NICHD created the Down Syndrome Working Group consisting of scientists from across the NIH to coordinate ongoing research and identify new research efforts. Working with the overall Down syndrome community, the Working Group in 2007 created and published the [NIH Research Plan on Down Syndrome](#), a comprehensive plan focused on genetic and neurobiological research relating to the cognitive effects associated with the condition. That plan was updated in December 2014 and includes a new section on "Down Syndrome and Aging," which recognizes the link between Alzheimer's disease and Down syndrome.

In an effort to foster communication and idea-sharing, the [Down Syndrome Consortium](#) was created in 2011 by NIH and consisting of individuals with Down syndrome and their families, national organizations interested in Down syndrome, and pediatric and other groups. One of its first activities was to create a Down syndrome registry, now called [DS-Connect®: The Down Syndrome Registry](#). Launched by NIH in 2014, the registry will store detailed information about people with Down syndrome who wish to participate in studies by doctors and scientists seeking to develop new and effective educational, behavioural, and drug treatments.

Additional public-private partnerships between NIH and the Down syndrome community are necessary to advance research and improve health outcomes for individuals with Down syndrome. For example, the NIH Research Plan on Down Syndrome recognizes the need for a centralized brain, organ and tissue biobank for Down syndrome so that researchers have a sufficient sources of biological samples to better understand the factors that underlie dementia and other health and cognitive conditions associated with Down syndrome. However, given the scarce public and private funding for Down syndrome biomedical and clinical research, such a centralized repository will be difficult to develop without a collaborative partnership with NIH similar to DS Connect.

Ask:

- **House Ask:** Urge House contact House Appropriations Subcommittee on Labor, HHS, Education and Related Agencies Chairman Tom Cole (R-OK) and Ranking Minority Member Rep. Rosa DeLauro (D-CT) to support these Down syndrome research priorities as part of legislation that appropriates annual funding for the National Institute of Health (NIH)

House Appropriations Subcommittee on Labor, HHS, Education and Related Agencies contacts:

Majority Staff John Bartrum at john.bartrum@mail.house.gov or (202) 225-3508

Minority Staff Siobhan Hulihan at siobhan.hulihan@mail.house.gov or (202) 225-3481

- **Senate Ask:** Urge Senators contact Senate Appropriations Subcommittee on Labor, HHS, Education and Related Agencies Chairman Blunt (R-MO) and Ranking Minority Leader Senator Murray (D-WA) to support these Down syndrome research priorities as part of legislation that appropriates annual funding for the National Institute of Health (NIH)

Senate Appropriations Subcommittee on Labor, HHS, Education and Related Agencies contacts:

Majority Staff Laura Friedel at laura_friedel@appro.senate.gov or (202) 224-9145

Minority Staff Lisa Bernhardt at lisa_bernhardt@appro.senate.gov or (202) 224-9145

