



Chairman Guthrie, Ranking Member Eshoo, and members of the Committee, thank you for the opportunity to testify at today's hearing.

My name is Michelle Sie Whitten, and I am the co-founder, President, and CEO of the Global Down Syndrome Foundation ("GLOBAL") and the mother of two, including a 20-year-old who happens to have Down syndrome. Like other parents of children with Down syndrome, our family considers Sophia a gift who has transformed our lives and the lives of those around her for the better.

From the moment I received the prenatal diagnosis that Sophia would have Down syndrome, I was motivated to learn as much as I could about the condition. I met with many clinical and scientific experts including then-Director of the National Institutes of Health (NIH), Dr. Elias Zerhouni, who informed me that Down syndrome was one of the least funded genetic conditions at NIH. He advised me that if I was to do one thing on behalf of the Down syndrome community, I should work to rebuild the pipeline for science and establish an academic home for Down syndrome medical research. I took his words to heart, and, with the support of my family, we did just that through GLOBAL and our Affiliates - Crnic Institute for Down Syndrome, Sie Center for Down Syndrome, and Alzheimer's & Cognition Center.

People with Down syndrome are born with an extra copy of chromosome 21. Researchers now know this extra chromosome results in a radically different disease profile compared to the general population and people with Down syndrome are highly predisposed to certain diseases and highly protected from others.

We now know people with Down syndrome are the largest population with a genetic predisposition to Alzheimer's disease but had been excluded from Alzheimer's clinical research for decades. By studying people with Down syndrome, we can not only help our loved ones with Down syndrome but also potentially millions of others who suffer from Alzheimer's.

The list of co-occurring diseases is considerable, I will just note two: 50 percent of babies born with Down syndrome have congenital heart defects and have a predisposition to autoimmune disorders such as celiac and autoimmune thyroid disease.

People with Down syndrome are also highly protected from developing solid tumors. For example, it is extraordinarily rare for someone with Down syndrome to have breast cancer or prostate cancer. On the other hand, they are highly predisposed to two blood cancers: acute megakaryoblastic leukemia and acute lymphoid leukemia.

In the 1980s, the average lifespan of a person with Down syndrome was 28 years old. I am pleased to report that with the renewed investment in Down syndrome research and medical care, the average lifespan today is 60 years. However, more needs to be done for people with Down syndrome in rural areas and within the Black population, where public health data indicates there is a substantially shorter life span as compared to White people with Down syndrome.



Down syndrome is a complex yet a manageable and treatable condition with research needs that involve almost each of NIH's 27 institutes and centers. Fortunately, bipartisan congressional champions recognized the magnitude of impact Down syndrome research could have on millions of Americans. Representatives Rosa DeLauro, Tom Cole, and Chair Rodgers have led the way in calling for increasing federal funding for such research and urging NIH to initiate a trans-NIH effort to study this unique disease spectrum. GLOBAL is truly grateful to have such fierce advocates championing the Down syndrome community and leaders who understood the value of therapeutic leverage.

There is no doubt that the creation of the INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome (INCLUDE) Project at NIH has been paramount in advancing our goals of increasing lifespan and improving health outcomes in children and adults with Down syndrome. Since FY18, INCLUDE has provided 556 unique awards from 18 different NIH institutes, with 11 institutes participating in Down syndrome research for the first time.

We can already point to advances in research and concrete outcomes resulting from INCLUDE:

1. Clinical Trials. Prior to INCLUDE there were few if any Down syndrome clinical trials and most clinical trials excluded people with Down syndrome from participating. In the six years since INCLUDE was established, there are now at least 12 clinical trials that include repurposed drugs to treat autoimmune diseases, Alzheimer's, cognition deficit, and Regression Disorder, and more.
2. Alzheimer's Research: INCLUDE has funded the Trial-Ready Cohort-Down Syndrome under the leadership of Dr. Mike Rafii who runs the Alzheimer's Therapeutic Research Institute at the Keck School of Medicine.
3. Data Coordinating Center: INCLUDE established the Data Coordinating Center that now has more than 9,000 participants, 4,000 whole genome sequences, hundreds of molecular data sets and other data sets that can launch the career of investigators interested in Down syndrome medical research.
4. New Investigator Training. INCLUDE has funded over 20 training grants helping us to ensure a pipeline of new and/or young investigators in the field.
5. Down Syndrome Registry. INCLUDE now houses the important Down syndrome registry, called DS Connects that was essentially directed by Congress into existence in December 2010 when GLOBAL and the NIH were required to organize the first-ever Down syndrome research conference at NIH looking at databases, registries, and biobanks.
6. Down Syndrome Clinical Cohort Coordinating Center. INCLUDE recently put out an RFA for the DS-4C with a centralized biobank. The establishment of such centers has been a GLOBAL goal since 2006 and to see it coming to fruition is very exciting and rewarding.

Through INCLUDE, we are seeing a renaissance for Down syndrome research and witnessing the great promise and tangible results from this project.



The “DeOndra Dixon INCLUDE Project Act of 2024” to authorize a trans-NIH model for Down syndrome would ensure crosscutting issues and opportunities are identified for researchers. This structure allows for multiple institutes to collaborate on a research plan and ensure institute coordination in terms of planning, funding, and sharing and disseminating research results. I want to extend our deepest appreciation for our sponsors paying tribute to one of GLOBAL’s most beloved and accomplished self-advocates who tragically passed away in 2020 at the age of 36. This legislation honors her and all individuals with Down syndrome.

I also want to acknowledge and express GLOBAL’s support for the Charlotte Woodward Organ Transplant Discrimination Prevention Act, which would prohibit healthcare providers, and matching entities, from discriminating against patients who need organ transplants solely based on disability. This bill highlights the importance of and need for auxiliary aids and services, a support system to assist with post-operative care, and the need for reasonable modifications to policies and procedures to make organ transplant systems and facilities more accessible to those with disabilities.

In addition, we want to share our support and appreciation for Chair Rodgers’ bill, the Protecting Health Care for All Patients Act, which bans ALL federal health programs from using discriminatory measures such as Quality-Adjusted Life Years to deny or restrict medical coverage for patients with Down syndrome or other conditions. It is important that this applies to all government health programs and not just Medicare.

While there is still much work to be done for us to close the gap on lifespan and health outcomes for those with Down syndrome as compared to those without Down syndrome, the momentum continues to build.

We are entering a new era when it comes to valuing people with Down syndrome and authorizing the INCLUDE Project ensures our dedicated researchers can continue their work to reduce health disparities, elongate life, and improve health outcomes not only for people with Down syndrome, but for everyone.

Thank you again for the opportunity to testify today and I look forward to your questions.