

Down Syndrome: Update on the State of the Science & Potential for Discoveries Across Other Major Diseases

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TESTIMONY

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Chairman Cole and Ranking Member DeLauro, thank you for convening today's hearing and for your work and leadership in significantly increasing federal support for biomedical research and efforts to improve the quality of life of Americans with Down syndrome.

Thank you Representatives McMorris Rodgers, Bustos and Sessions for all you have done and continue to do for our children and adults with Down syndrome. Representatives McMorris Rodgers and Sessions have been great mentors to me and advocates for my organization's work.

My name is Michelle Sie Whitten. First and foremost I am a daughter, a wife and a mother. I am the daughter of two immigrants, my mother is from Italy and my father is from China. And I am the mother of two fabulous children – Sophia who is 14 and happens to have Down syndrome and Patrick who is a typical 11 year old. They are all here in the audience today with my husband and his mom and sister from England, and I thank them for allowing me to work hard every day to improve the future not just for Sophia but for Cole, Alex and millions more with Down syndrome.

When I was pregnant with Sophia I had an amnio. The genetic counselor told me my baby would die by three and essentially pressured me to terminate. My husband and I did some soul searching and continued with the pregnancy and we never looked back. We consider Sophia a gift who has enriched our lives and all those around her.

During my pregnancy I discovered the lifespan of a person with Down syndrome was not three years but fifty. I also discovered there was little or no clinical or biomedical research addressing health outcomes for people with Down syndrome.

Shortly after I gave birth I found myself in Bethesda meeting with the then Director of the National Institute of Health (NIH), Dr. Elias Zerhouni.

It was Dr. Zerhouni who pointed out to me that Down syndrome was one of the least funded genetic conditions at the NIH, and who told me "If you do just one thing – establish an academic home for Down syndrome and rebuild the pipeline for science." The science needs to be there to change this status quo.

We did just that. In 2008, my family and I organized a Down syndrome scientific summit and the conclusion was twofold: First, shock that there was not more funding for Down syndrome research. Second, conviction that research would not only help the 300K+ people with Down syndrome in the US but that it could help millions of others who suffer from diseases to which people with Down syndrome are highly predisposed to, what my father calls “Therapeutic Leverage.”

For example: 100% will have the brain pathology of Alzheimer’s disease by their 40s, up to 30% have an autoimmune disorder such as Celiac disease or Type 1 Diabetes, people with Down syndrome are 50 times more likely to develop leukemia and 500 times more likely to develop acute megakaryoblastic leukemia (AMLK). And yet, it is extraordinarily rare for a person with Down syndrome to suffer from solid tumor cancers (e.g. breast cancer, prostate cancer) or heart attacks.

Importantly, these medical conditions that people with Down syndrome are either predisposed to or protected from cause more than 50% of deaths in the United States and other developed countries.

Based on this knowledge, we established the Global Down Syndrome Foundation in 2009. Our mission is to significantly improve the lives of people with Down syndrome through research and medical care. We collaborate with several other national Down syndrome organizations doing excellent work in other areas and they support us taking a lead on these issues.

Global has worked tirelessly and as good partners with Congress and the NIH to stimulate Down syndrome research funding. Shortly after we incorporated, Global worked with Representative Sessions to help start the Congressional Caucus, today a bicameral task force, with former U.S. Representative Patrick Kennedy.

When the NIH had additional funds from the American Recovery and Reinvestment Act (ARRA), Global worked with renowned Johns Hopkins Down syndrome researcher Roger Reeves and 18 Down syndrome investigators to annotate \$8 million of Down syndrome grants that were not fully funded or not funded at all that could be considered. Unfortunately these grants did not receive any of the ARRA funding.

We have reached across the aisle and worked with U.S. Senators Tom Harkin, the late Arlen Specter, and Richard Shelby. Today we are privileged to have close friends and allies like U.S. Representatives Cathy McMorris Rodgers, Rosa DeLauro and Pete Sessions and many other colleagues who have supported language in the Labor Health and Human Services Appropriations legislation reflecting the need to correct the disparity of funding for Down syndrome research.

In December of 2010 we jointly organized the first ever Down syndrome conference held by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

focused on a national registry and biobanks. The conference findings were published in 2011 and were a catalyst to NICHD's Down syndrome registry called DS-Connect.

Most importantly, we have established a strong pipeline of excellent science. Today, I am proud to report that a lack of good science or lack of investigators submitting proposals can no longer be a rationale for the disparity of funding for Down syndrome research. Today, Global's gross revenue is approximately \$8 million with the majority of our proceeds going to research, and our programs reach a quantifiable 20,000 people with Down syndrome in the US including medical care to patients from 28 states and 7 countries. We have 38 labs and over 140 scientists working on Down syndrome with key focuses on Alzheimer's, autoimmune disease and cancer.

Today, we are ready to work with a trans-NIH platform that builds on amazing breakthrough that allows us to recast Down syndrome as an immune system disorder. Our scientists discovered that the interferon pathway, the pathway that is lit up only when fighting a virus or infection in typical people, is lit up 24/7 in people with Down syndrome from the time they are born until they die. This tax on the immune system can help explain the co-occurrence of Alzheimer's and autoimmune disease and protection from solid tumor cancers. Most hopeful, there are FDA-approved drugs that can bring down those high levels of interferon activity.

The science stands ready, but now we need our colleagues at the NIH to think in new ways with us to leverage these amazing breakthroughs. Consider with me how this can be done.

If we take the key comorbidities associated with Down syndrome, then it would follow that at least ten NIH institutes should be key stakeholders in Down syndrome research – those 10 are listed in your handout. People with Down syndrome stand ready to serve as a lens through which cancers, Alzheimer's, autoimmune diseases and many other conditions can be studied and understood. The community stands ready to become actively engaged as partners in this extraordinary scientific endeavor. While NICHD will always be the natural home for Down syndrome research, this research cannot be silo'd at the NICHD where they are tasked to fund hundreds of diseases and programs with a very small budget.

On behalf of my daughter's future, our constituents with Down syndrome and their families, and others that stand to gain from our science, we hope that Congress can help effectuate a trans-NIH approach to Down syndrome research consistent with the 21st Century Cures law, and that our mutual desire for NIH to engage in outside of the box thinking and the incorporation of new funding into this research is fulfilled.

This new endeavor will fundamentally change the unacceptable trajectory for Down syndrome research. Let me give you a picture of this trajectory.

Despite our advocacy and advancements, there has been negative to flat funding for Down syndrome research. There is also a significant disparity as compared to other developmental

conditions or comparable disorders. My written testimony includes an NIH authored budget chart that illustrates this challenge.

As you can see from this chart, despite being the leading cause of developmental delay in the U.S. and the world, Down syndrome is one of the least funded genetic conditions by the NIH. From 2001 to 2006 NIH annual funding for Down syndrome research plummeted from \$29 million to \$14 million despite significant growth of the NIH budget during this time.

From 2001 to 2017 IF Down syndrome had just increased and decreased even at the low amount with the ups and downs of the NIH, the funding would have been \$744 million – more than double the actual \$356 million cumulative for those 19 years.

I will leave you with these final numbers:

1. Based on the CDC, There are over 300,000 people estimated with DS living in the US today
2. Live births have increased from 1 in 1,000 in 2002 to 1 in 691 today
3. The lifespan of a person with DS has more than doubled to 60 years today up from 28 years in the 1980s
4. With increased live births and a doubling of lifespan, there will be a relative population explosion of people with Down syndrome over the next several decades

To be clear the number of people with Down syndrome is growing and the need is getting larger and not smaller.

Our children and adults with Down syndrome who are American citizens deserve to know that there is research funding and medical care available to them that allows them to reach their true potential.

Thank you for caring about the future of this special population and allowing me to testify at this milestone hearing.