

**FY 2019 Public Witnesses Hearing for the  
House Subcommittee on Labor, Health and Human Services, Education and Related Agencies  
Thursday, April 26, 2018**

Testimony from Michelle Sie Whitten, MA, Founder, President & CEO, Global Down Syndrome Foundation

Chairman Cole and Ranking Member DeLauro, thank you for convening today's hearing and inviting the Global Down Syndrome Foundation (Global) to present along with so many important causes represented by impassioned and dedicated professionals and advocates. For Global and the Down syndrome community, our hope is that this esteemed committee can help us to ensure that the National Institutes of Health (NIH) will take advantage of the generous \$3 billion in new funding included in the FY2018 Consolidated Appropriations Act and make major investments in Down syndrome research. In particular, we hope that the funding of research benefitting people with Down syndrome will increase significantly from FY2017 to FY2018 and then again from FY2018 to FY2019, and that the increase will be seen in both the current pipeline of Down syndrome research and the new trans-NIH Down syndrome research initiative.

As the mother of Sophia, soon to be 15, who happens to have Down syndrome, and as the President and CEO of Global, I am truly honored to be here and I hope that my testimony may help ensure that our history will not be forgotten and that with your support, research benefitting people with Down syndrome at the NIH will not decline but rather continue to grow. This will not only help the estimated 300,000+ Americans with Down syndrome but millions of Americans who suffer from diseases that can be unlocked or even cured through Down syndrome research.

Global has been working hard with the Down syndrome community, scientists, the NIH, and Congress for many years to address the troubling disparity of funding for Down syndrome research. In October of last year, we were thrilled to have the opportunity to present here, at the first-ever Congressional Appropriations hearing on Down syndrome research. This was an important milestone for raising awareness of the opportunities presented by Down syndrome research and brought us much hope for working with Congress to advance this research in the future.

For this hope, and for the promise I believe it will deliver, I am deeply and forever grateful to every single member on this subcommittee who, in a bipartisan fashion, continues to grow the NIH budget and understands the urgent need to make significant new investments in Down syndrome research overall and to fully fund the trans-NIH initiative on Down syndrome recommended by this subcommittee in the FY 2018 Consolidated Appropriations Act. We are also indebted to the leadership of Representatives McMorris Rodgers, Bustos, Sessions and Coffman for all they have done and continue to do for our children and adults with Down syndrome.

Despite being the leading cause of developmental delay in the U.S. and in the world, Down syndrome is one of the least funded genetic conditions by our NIH and has been for the last two decades. Beginning in 2001 when Down syndrome research funding was at \$29 million (or 0.14% of the NIH budget), there was a precipitous decline in funding for this research during double digit growth of the overall NIH budget. As a result, annual funding for Down syndrome research fell to \$14 million in 2006 and continued to flat line and even decrease until 2017

when the funding total was \$28 million (or 0.09%% of the total NIH budget). *You can see the numbers in a handout we have provided.*

Today I will present two compelling reasons why the need for increased funding for Down syndrome research is so urgent. First, we should help, not hurt, people with Down syndrome. Even as compared to other developmental disabilities, Down syndrome research is woefully underfunded. People with this condition deserve improved medical care that is specific to their radically different disease spectrum caused by having three copies of chromosome 21 instead of two. Because of lack of research, our children and adults suffer needlessly, we are still unclear about several key comorbidities, and we have no medical care guidelines for adults with Down syndrome. Clearly, good health improves the reality of how our self-advocate, Frank Stephens, defines his own life: “a life worth Living.”

And unlike Iceland or Denmark, where there is a call to be “Down syndrome-free” by 2030, there is actually a mini-population explosion of people with Down syndrome in the U.S. that cannot be ignored. (1) Based on CDC numbers, there are over 300,000 people estimated with DS in the US. (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. With increased live births and a doubling of lifespan, we happily don’t have to be mathematicians to know there is a relative population explosion of people with Down syndrome that will continue over the next several decades

Second, people with Down syndrome are either predisposed to or protected from diseases that cause more than 50% of deaths in the United States and other developed countries. 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 (AMKL). 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. Clearly, by involving people with Down syndrome in research, we stand to improve their lives and tens of millions of others.

Thanks to the leadership of this subcommittee, the FY 2018 Consolidated Appropriations Act directs the NIH to establish a new trans-NIH research initiative supported by an increase in funding that will study the connection between Trisomy 21 and other co-occurring conditions. We believe this will help address the research challenges that our community has faced for the last two decades. At the FY2019 NIH budget hearing held two weeks ago, our community was pleased to see that many NIH institutes are embracing this initiative as a way to break down silos, share information, and focus on precision-medicine – all key goals at the NIH.

As presented in the October hearing last year, Global and other research organizations have established a small, private mechanism for funding excellent science. Our scientists at Global's affiliate, the Linda Crnic Institute for Down Syndrome, recently made a transformative discovery whereby we are able to recast Down syndrome as an immune system disorder. Three new papers in high-impact journals will be published in this quarter alone on the subject. We 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 taxes the immune system and can help explain the co-occurrence of

Down syndrome with Alzheimer's and autoimmune disease and protection from solid tumor cancers. This research also holds promise because there are FDA-approved drugs that can bring down those high levels of interferon activity.

The Down syndrome research community (including researchers at Johns Hopkins, University of San Diego, University of CA Irvine, Columbia University, Northwestern University), stand ready to advance this science. We are eager to work with our colleagues at the NIH to leverage these amazing breakthroughs and to help the research get to the next level.

Global and the Down syndrome research community hope to work with this subcommittee and the NIH to ensure that funding for Down syndrome research continues to grow in FY 2019. We also hope to work together to further define the trans-NIH initiative including what institutes will be involved, what grant or research mechanisms will be utilized, and how the study of immune system dysregulation can help unlock the mysteries of other diseases. Please have no doubt that Global will use its utmost ability, resources, and hard work to help deliver on this important and historic initiative!

On behalf of my daughter's future, our constituents with Down syndrome and their families, our national and local colleagues in the Down syndrome field, and others that stand to gain from our science, we thank you for listening, caring and ACTING! Our children and adults with Down syndrome who are American citizens deserve to know that there is research funding and medical care available 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 important hearing.



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**TESTIMONY ATTACHMENT**

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<b>Year</b>	<b>NIH Actual Total Obligations by Budget Mechanism (In Millions, Rounded)</b>	<b>CF Research Funding (Dollars in Millions)</b>	<b>Fragile X Research Funding (Dollars in Millions)</b>	<b>MS Research Funding (Dollars in Millions)</b>	<b>Autism Research Funding (Dollars in Millions)</b>	<b>DS Research Funding (Dollars in Millions)</b>	<b>DS Research Funding to NIH Budget (Rounded)</b>
2000	17,814					27	0.0015
2001	20,513					29	0.0014
2002	23,188					28	0.0012
2003	26,740					23	0.0009
2004	28,100					19	0.0007
2005	28,626					15	0.0005
2006	28,533					14	0.0005
2007	29,034					16	0.0006
2008	29,320	90	26	169	118	17	0.0006
2009	30,207	86	27	137	132	18	0.0006
2010	31,036	86	25	133	160	22	0.0007
2011	30,630	79	29	122	169	20	0.0007
2012	30,802	86	27	115	192	20	0.0006
2013	29,137	78	30	112	186	18	0.0006
2014	30,019	77	36	102	188	18	0.0006
2015	30,293	80	38	94	208	24	0.0008
2016	32,259	89	44	97	232	27	0.0008
2017 (Est.)	32,259	91	46	101	243	28	0.0009

**There are at least 10 institutes at NIH whereby Down syndrome research could be a directly relevant discovery accelerator:**

1. Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)
2. National Institute on Aging (NIA)
3. National Institute of Allergy and Infectious Diseases (NIAID)
4. National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)
5. National Institute of Diabetes and Digestive and Kidney Diseases (NIDDKD)
6. National Heart, Lung and Blood Institute (NHLBI)
7. National Eye Institute (NEI)
8. National Center for Advancing Translational Sciences (NCATS)
9. National Institute of Neurological Disorders and Stroke (NINDS)
10. National Cancer Institute (NCI)

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