Testimony on behalf of the American Society of Human Genetics, Submitted to the House Appropriations Subcommittee on Labor, Health, and Human Services, Education, and Related Agencies, Anthony Wynshaw-Boris, MD, PhD, President-Elect

The American Society of Human Genetics (ASHG) thanks the Subcommittee for its continued support and leadership in funding the National Institutes of Health (NIH). The $2 billion increase provided for FY 2019 reinforced our nation's commitment to the health and well-being of all Americans by investing in biomedical research and scientific innovation. **ASHG urges the Subcommittee to appropriate $41.6 billion for the NIH in FY 2020.**

My name is Tony Wynshaw-Boris. I am a professor and chair of the Department of Genetics and Genome Sciences at Case Western Reserve University. My laboratory studies the biology, specifically the genetics, of the development and function of the brain.

Transforming Human Health with Genomics

The transformative impact of genetics research for fundamental scientific knowledge and health applications is profound. Thanks to decades of sustained investment in basic and applied research, we are moving rapidly into an exciting new era of genetic investigation, novel application, diagnosis, and treatment. Due to NIH funding of the development of innovative sequencing technologies, it is now possible to sequence an individual's genome rapidly and cost-effectively. Remarkably, whereas fifteen years ago it cost tens of millions of dollars to sequence a human genome, today it can be done for around a thousand dollars in about a day[1]. From diagnosis to prognosis and treatment,

this now-affordable and accessible data about genetic risk and resiliency for individuals and populations is ushering in a new era of precision medicine. Genetic and genomic approaches are driving novel discoveries across the spectrum of biomedical research. These advances are a central component of the research agenda of most NIH Institutes and Centers. For FY 2017, $3.3 billion of NIH's $34.2 billion budget was spent on human genome-associated research projects[2].

To realize the benefits of genetics and genomics research for all people, NIH-funded basic, translational, and clinical genetics research must continue if we are to fuel further progress. An illustrative example is cancer. Today, researchers have discovered more than 50 hereditary cancer syndromes[3]. Screening for cancer-causing mutations as a risk indicator, such as mutations in the genes BRCA1 and BRCA2, allows for increased monitoring and early intervention strategies. Sequencing tumors for changes that may identify effective drugs for that particular tumor can be used to guide treatment and improve prognosis. Finally, the recent approval by the Food and Drug Administration (FDA) of two gene therapies, Kymriah and Yescarta for treatment of acute lymphoblastic leukemia and B cell lymphoma respectively, marks an important breakthrough in cancer treatment options[4]. It is essential to continue support and expand research to develop targeted therapies for the millions of patients across our country affected by numerous other cancers.

Another example of progress and future promise can be found with opioid addiction -- a national crisis with increasing burden on public health. NIH-supported genomics research is revealing new insights into the complex causes of addiction. Genome-wide association studies have linked numerous genetic variants to increased

risk of opioid dependence, vulnerability to pain, and subsequent addiction[5]. We are also learning more about how genetics affects how the body breaks down opioids and other drugs. Clinics and hospitals have started using this information to guide pain management options[6].

Other studies are focusing on the link between genomics and infectious diseases. Scientists recently discovered that a genetic variant in a gene for an antiviral protein is related to the severity of influenza[7]. An ongoing clinical trial is testing how genetic variants in flu vaccine recipients affects their protection from the flu[8].

The discovery of CRISPR-Cas9 as a gene-editing technique in research laboratories holds promise as an exciting new approach to treating diseases. Gene-editing methods can precisely modify a DNA sequence in a cell to correct the gene variant that is causing disease. Multiple clinical trials are underway testing gene-editing strategies as treatments and even cures for diseases such as cancer, sickle cell anemia, and inherited childhood blindness[9].

The impact of NIH-funded activities extends beyond public health

Return on federal funding for NIH can also be quantified by its economic impact. In FY 2017, approximately 83% of the budget was allocated to more than 2,500 universities and institutions across every state, supported over 400,000 jobs and stimulated $69 billion in economic activity[10]. An example of this is Yescarta, noted above, which was a key product in the success of Kite Pharma, which was acquired for nearly $12 billion by another successful American company, Gilead Sciences. Likewise, we see a remarkable effect in the genomics research field. The Human Genome Project alone,

and subsequent research has been shown to yield a total economic output of roughly $1 trillion and $55 billion in tax revenues over a 24-year span[11]. Each dollar invested in genomics research contributed $65 to the U.S. economy.

Given NIH's significant impact in many facets of the lives of our people and the growth and competitiveness of our economy, ASHG joins the Federation of American Societies for Experimental Biology (FASEB), and the Ad Hoc Group for Medical Research in recommending a $41.6 billion budget for NIH for FY 2020.

The American Society of Human Genetics (ASHG), founded in 1948, is the primary professional membership organization for human genetics specialists worldwide. The Society's nearly 8,000 members include researchers, academicians, clinicians, laboratory practice professionals, genetic counselors, nurses and others who have a special interest in the field of human genetics.

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[1] https://www.genome.gov/10000008/budget-and-financial-information/

[2] https://report.nih.gov/categorical_spending.aspx

[3] https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet

[4] https://www.cancer.gov/news-events/cancer-currents-blog/2018/tisagenlecleucel-fda-lymphoma

[5] Hu, R. *et al*. 2018. ANCO-GeneDB: annotations and comprehensive analysis of candidate genes for alcohol, nicotine, cocaine and opioid dependence. *Database* 2018.

[6] Cavallari, L.H. *et al*. 2019. Multi-site investigation of strategies for the clinical implementation of CYP2D6 genotyping to guide drug prescribing. *Genetics in Medicine* 2019.

[7] Allen, E.K. *et al*. 2017. SNP-mediated disruption of CTCF binding at the IFITM3 promoter is associated with risk of severe influenza in humans. *Nature Medicine* 23, 975-983.

[8] http://med.stanford.edu/vaccines/clinical_trials.html

[9] https://clinicaltrials.gov/

[10] http://www.unitedformedicalresearch.com/advocacy_reports/nihs-role-in-sustaining-the-u-s-economy2018-update/

[11] https://web.ornl.gov/sci/techresources/Human_Genome/publicat/2013BattelleReportImpact-of-Genomics-on-the-US-Economy.pdf