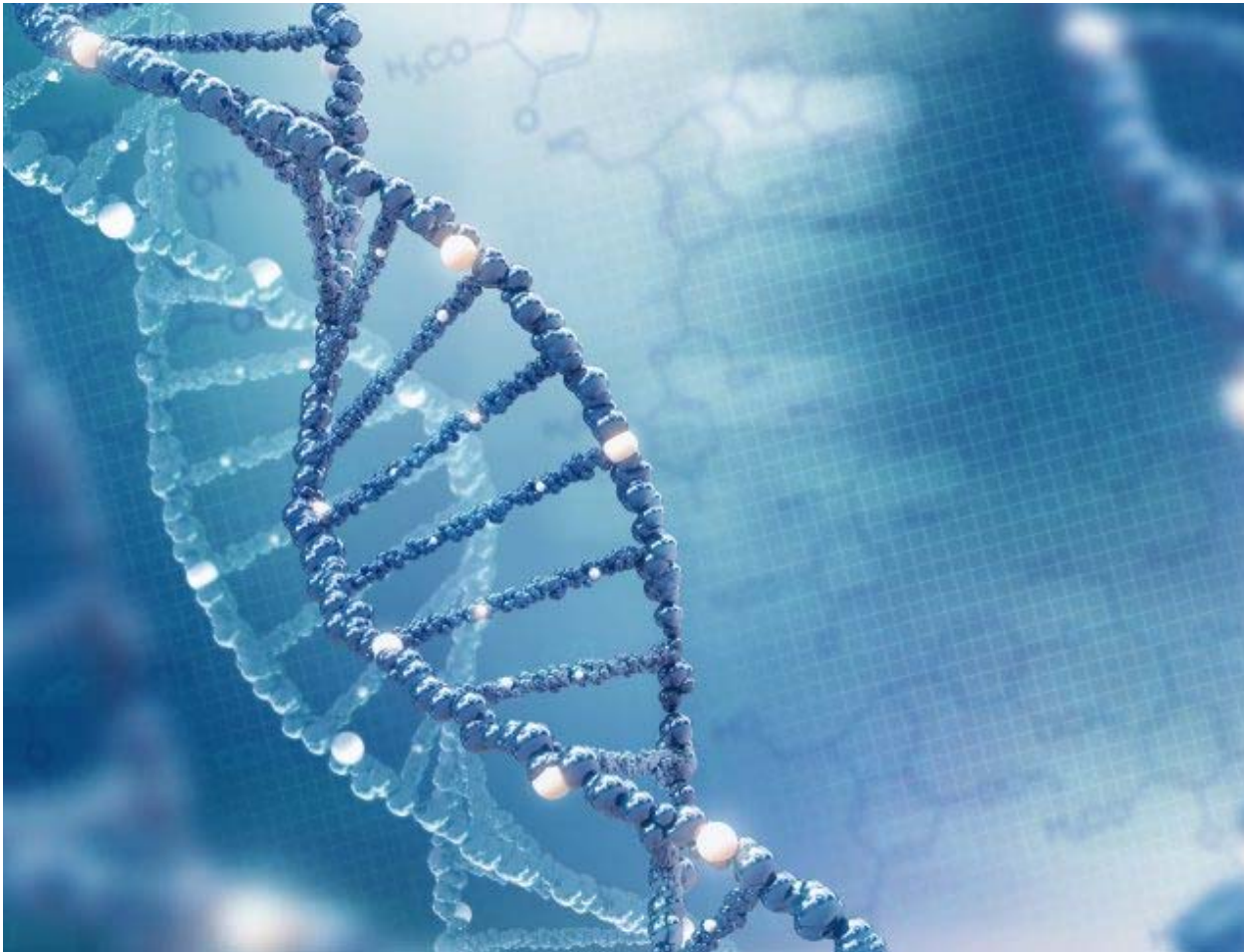


NHGRI Funds Two Centers as Part of Human Genome Reference Program

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The NIH's National Human Genome Research Institute (NHGRI) has awarded a total \$29.5 million in grants over five years to two centers as a part of a new Human Genome Reference Program (HGRP) that will aim to advance the most comprehensive reference sequence of the human genome. [Zffoto / Getty Images/iStockphoto]

The NIH's National Human Genome Research Institute (NHGRI) said today it will fund two centers as a part of a new Human Genome Reference Program (HGRP) that will aim to advance the most comprehensive reference sequence of the human genome, through new grants totaling \$29.5 million over five years.

The NHGRI has awarded \$2.5 million per year for five years to Washington University in St. Louis (WashU), University of California, Santa Cruz (UCSC) and European Bioinformatics Institute (EBI), which will coordinate with the National Center for Biotechnology Information to form the WashU-UCSC-EBI Human

Genome Reference Center. The center is intended to provide a next-generation reference sequence of the human genome for, as well as support interactions with the community of genomics researchers.

UCSC and WashU are also part of an international consortium to which NHGRI awarded approximately \$3.5 million per year over five years toward a Human Reference Genome Sequencing Center that aims to sequence up to 350 additional diverse human genomes, using advanced technologies designed to incorporate quality sequences that are more broadly representative of the population. Other institutions within the consortium include University of Washington School of Medicine, The Rockefeller University, Mt. Sinai, Harvard University, Broad Institute, Coriell Institute for Medical Research, McGill University, University of Cambridge and the Max Planck Institute.

“We finally have the technology and methods to go after the parts of the human genome that were beyond our reach 20 years ago,” Evan Eichler, PhD, professor of genome science at University of Washington and a Howard Hughes Medical Institute investigator, said in a UW statement. “It’s an exciting time for human genetic implications for improved variant discovery associated with disease.”

Eichler’s group studies structural variation in the human genome and on genomic instability, as well as for contributions to comparative DNA sequencing and autogenetics. His team will focus on more difficult genome regions that vary among individuals and will use advances in highly accurate long-read sequencing to characterize those areas, UW stated.

Plans for a “Pan-Genome”

Over time, the two centers will work with international collaborators to develop a “pan-genome” or multi-genome reference sequence that is as universal and complete as possible. This more-complete reference sequence will enable analysis of any human DNA sequence by reflecting all human diversity, according to NHGRI.

“It has grown more and more important to have a high-quality, highly usable human genome reference sequence that represents the diversity of human populations. The proposed improvements will serve the growing basic and clinical genomics research communities by helping them interpret both research and clinical genome sequences,” Adam Felsenfeld, Ph.D., NHGRI program director in the

Division of Genome Sciences, said in a statement.

Just as people use a reference photo to assemble pieces into a jigsaw puzzle, researchers plan to use the reference genome sequence to assemble genome sequences from individuals. The HGRP will enable researchers to find disease-causing variants and specify their genomic locations with markedly increased accuracy, according to NHGRI.

Grant awards are subject to the availability of funds, the NHGRI cautioned.

Earlier this year, the NHGRI said it also expected to award (https://www.genome.gov/sites/default/files/genome-old/pages/Research/Sequencing/HumanGenomeReferenceProgram/HGRP_Webinar_FAQ.pdf) two to four awards for R&D for Reference Representations at \$1.25 million per year (total costs for awards combined); and two to four awards for the Notice for Comprehensive Human Genome Sequencing Methodologies at \$1.5M /year.

