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Scientists Apply Successful Single Molecule Sequencing and *de novo* Genome Assembly to a Parasitic Worm that Infects Human Eyes and Skin

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Investigators at the Institute for Genome Sciences (IGS) at the University of Maryland School of Medicine and the Laboratory of Parasitic Diseases at the National Institute of Allergy and Infectious Diseases (NIAID) at the National Institutes of Health (NIH) used the long-read, single-molecule Pacific Biosciences platform for the successful genome sequencing and *de novo* assembly of *Loa loa* roundworms from a clinical sample. Their research, which generated the most complete genome sequence of a filarial nematode produced to date, provides a more comprehensive reference genome for this parasite in the hopes of developing better molecular diagnostics to decrease morbidity from filarial nematodes. Their findings appear in today's issue of *BMC Genomics*.

More than 20% of the world's population is at risk for infection by filarial nematodes and more than 180 million people worldwide are already infected. The *Loa loa* roundworm, which is the causative agent of loiasis ("African eye worm"), enters the human body through the bite of a deer fly. It lives beneath the skin of its host, often undetected for years, where it can grow to be several centimeters in length.

Because these roundworms have multiple hosts and cannot be reared or easily manipulated in the lab, genome sequencing provides us with much need information about these filarial nematodes, which are poorly understood organisms that cause several neglected tropical diseases including filariasis, river blindness, and loiasis. Yet there are numerous challenges in sequencing filarial nematode genomes, including obtaining the necessary clinical samples, and assembling these large genomes from genetically heterogeneous DNA that is highly repetitive and AT-rich. Prior to the advent of lower cost long-read sequencing platforms like the Pacific Biosciences RS II, efforts to sequence these genomes have suffered from the limitations of short-read technologies and high costs. Here, these scientists demonstrate that the technology reconstructs more complete genomes for a fraction of the previous costs from small amounts of heterogeneous DNA taken from a single clinical sample.

"Recent improvements in long-read, single-molecule sequencing have enabled more economical sequencing and improved genome assembly for previously difficult to sequence clinical samples. To our knowledge, this study represents the largest and most complete genome of an uncultured clinical specimen successfully sequenced and assembled using this technology," says Luke Tallon, Scientific Director of the IGS Genomics Resource Center, whose team performed the sequencing and assembly.

"We are excited to be able to rapidly produce such high quality genomes at a fraction of previous costs enabling better and more comprehensive studies of important understudied groups of organisms. We look forward to applying this approach to other metazoan genomes," says Julie Dunning Hotopp, Ph.D., Associate Professor of Microbiology and Immunology at the Institute for

Genome Sciences (IGS) at the University of Maryland School of Medicine and senior author on the paper.

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About the Institute for Genome Sciences

The Institute for Genome Sciences (IGS) is an international research center within the University of Maryland School of Medicine. Comprised of an interdisciplinary, multidepartment team of investigators, the Institute uses the powerful tools of genomics and bioinformatics to understand genome function in health and disease, to study molecular and cellular networks in a variety of model systems, and to generate data and bioinformatics resources of value to the international scientific community. www.igs.umaryland.edu

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