

| New mammalian reference genome to aids in identification of genetic variants for human health

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BY JOHN EDWARDS ON DECEMBER 19, 2020

SCIENCE

The Rhesus monkey is the most widely studied nonhuman primate in biomedical research. A genome sequence project for this species led by researchers at Baylor College of Medicine, the University of Missouri and the University of Washington has established a new framework for studying this important primate. Published in the journal Science, the research has created a new reference genome and identified more than 85 million genetic variants in the rhesus monkey – the largest database of genetic variation to date for a nonhuman primate species.

“This is a big step forward in the amount of information we have about genetic variation in the rhesus monkey,” said Dr. Jeffrey Rogers, associate professor at the Human Genome Sequencing Center and Department of Molecular and Human Genetics at Baylor and one of the study’s corresponding authors. “We actually identified thousands of new mutations in the research animal population. Now colleagues around the country who are studying different aspects of health and disease with rhesus macaques can begin to use that information.”

Dr. Wesley C. Warren of the University of Missouri and Dr. Evan Eichler of the University of Washington, both corresponding authors on the study, used a combination of several advanced technologies to significantly improve the rhesus monkey reference genome first created in 2007. The new reference genome, published today, provides a more complete and accurate picture of the rhesus monkey DNA sequence. This improvement can support more sophisticated and detailed analyses of fundamental questions in molecular genetics, cell biology and physiology.

“When we coupled this higher-quality genome reference with broader sequencing of expressed genes, our team discovered gene structures unique to the rhesus monkey, as well as specific repeat sequences that are subject to deletions, suggesting that evolutionary

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competition to suppress the spread of these elements continues to this day,” Warren, a professor at the University of Missouri’s Bond Life Sciences Center, said.

Scientists at Baylor’s Human Genome Sequencing Center sequenced the genomes of 853 rhesus macaques from research institutions across the country and compared them to the new reference genome. The genome analysis showed that rhesus macaques have more genetic variants per individual than humans. Among the millions of genetic variants identified, the researchers found several deleterious mutations in genes known to cause genetic disorders in humans, including autism, inherited blindness and several others.

“We can find naturally occurring models of genetic disorders by studying the rhesus monkey population,” Rogers said. “We will find animals that naturally carry interesting and useful genetic mutations that can help us understand genetic variation and susceptibility to disease in humans. Rhesus monkeys are also intensively studied by primatologists and evolutionary biologists, so this new reference genome will also provide new insights into the evolution of non-human primate and human genomes.”

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