

Sea sponge helps scientists unravel 700-million-year-old mystery of evolution

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Dr Emily Wong and Associate Professor Mathias Francois. Credit: Victor Chang Cardiac Research Institute

A jelly-like sea sponge has helped shed light on an elusive part of the human genome, with implications for biomedical research and healthcare.



Australian scientists have found that humans, and most likely the entire animal kingdom, share important genetic mechanisms with a jelly-like sea sponge that comes from the Great Barrier Reef.

Published in *Science* today, the research reveals some elements of the human <u>genome</u>—an organism's complete set of DNA—are functioning in the same way as the prehistoric sea sponge. This mechanism—which drives gene expression, key to species diversity across the animal kingdom—has therefore been preserved across 700 million years of evolution.

UNSW scientist Dr. Emily Wong, who is based at the Victor Chang Cardiac Research Institute, says unravelling a mystery of this magnitude is significant.

"This is a fundamental discovery in evolution and the understanding of genetic diseases, which we never imagined was possible. It was such a far-fetched idea to begin with, but we had nothing to lose so we went for it," Dr. Wong says.

"We collected sea sponge samples from the Great Barrier Reef, near Herron Island. At the University of Queensland, we extracted DNA samples from the sea sponge and injected it into a single cell from a zebrafish embryo. Without harming the zebrafish, we then repeated the process at the Victor Chang Cardiac Research Institute with hundreds of embryos, inserting small DNA samples from humans and mice as well."

Dr. Wong says despite a lack of similarity between the sponge and humans due to millions of years of evolution, the team identified a similar set of genomic instructions that controls gene expression in both organisms.

"We were blown away by the results," Dr. Wong says.



According to scientists, the sections of DNA that are responsible for controlling <u>gene expression</u> are notoriously difficult to find, study and understand. Even though they make up a significant part of the human genome, researchers are only starting to understand this genetic "dark matter".

"We are interested in an important class of these regions called 'enhancers'," Dr. Wong says.

"Trying to find these regions based on the <u>genome sequence</u> alone is like looking for a light switch in a pitch-black room. And that's why, up to this point, there has not been a single example of a DNA sequence enhancer that has been found to be conserved across the animal kingdom.

"We are still a long way from a clear understanding of how DNA precisely shapes morphology in health and disease but our work is an important step in that direction."

Working alongside Dr. Wong is her husband and co-senior author on the paper, Associate Professor Mathias Francois from the Centenary Institute.

"This work is incredibly exciting as it allows us to better 'read' and understand the human genome, which is an incredibly complex and everchanging instruction manual of life," says A/Prof. Francois. "The team focused on an ancient gene that is important in our nervous system but which also gave rise to a gene critical in heart development."

The findings, he says, will also drive biomedical research and future healthcare benefits. "Being able to better interpret the human genome aids our understanding of human processes, including disease and disorders, many of which have a genetic basis. The more we know about



how our genes are wired, the better we are able to develop new treatments for diseases."

Marcel Dinger, Professor and Head of UNSW Science's School of Biotechnology and Biomolecular Sciences (BABS), said there was so much about the information stored in the genome that we still don't understand fully. "This study is as an important step towards decoding life's programming language—the new knowledge it presents will help inform future research across the medical, technology and life sciences fields. It's terrific to see such important research recognised by one of the world's most prestigious scientific journals—this really supports our ambition to be the best school of molecular biosciences in Australia."

More information: E.S. Wong el al., "Deep conservation of the enhancer regulatory code in animals," *Science* (2020). <u>science.sciencemag.org/cgi/doi ... 1126/science.aax8137</u>

N. Harmston el al., "Regulation in common: Sponge to zebrafish," *Science* (2020). <u>science.sciencemag.org/cgi/doi ... 1126/science.abe9317</u>

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