

Mutations that matter

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(Medical Xpress)—Identifying the genetic source of a specific trait can be a little like finding a needle in a field full of haystacks. University of Dayton biologist Thomas Williams is working to shrink the number of haystacks.

In a new study, published by *PLoS Genetics*, Williams and his lab team were able to single out a specific sequence in the genetic code of a fruit fly where changes to the code resulted in a different pigmentation pattern between flies of the same and related species. So out of the whole fly [genome](#)—more than 15,000 genes and even more genetic "switches"—[mutations](#) causing similar pigmentation changes occurred in the same switch.

"Scientists have mapped the genomes of humans and several animals, yet our understanding of how that information is stored and how it produces traits is woefully inadequate," Williams said. "There are literally millions of places where changes occur, but only thousands of places where it matters."

By studying the fruit fly as a model, Williams hopes to identify prime genetic "real estate"—those locations in the genome where mutations are most likely to have a significant impact, either positive or negative. Other scientists can then use that knowledge to focus studies on similar places in the [human genetic code](#), possibly identifying sections of the code where changes can pose [health problems](#) or [genetic risk factors](#).

"We want to save time and money," Williams said. "We want to help

others distinguish the signals from the noise."

It is common knowledge in the scientific community that genomes include the information for genes, which make a product that does something, Williams said. Less well known is that information is encoded in sequences that act as "switches" to tell genes when—and in which cells—to make their product.

The location for mutations occurs randomly in genomes. Many of these mutations are neutral, with no measurable effect on encoded information. Some mutations degrade information, leading to disease or disease risk. And some mutations modify information in useful ways.

"The great challenge for the era is to determine which mutations are neutral, injurious or beneficial," Williams said. "This difficulty is especially challenging for so-called genetic 'switches.' "

The paper, titled "Recurrent Modification of a Conserved Cis-Regulatory Element Underlies Fruit Fly Pigmentation Diversity," was published Aug. 29 by *PLoS Genetics*.

More information: www.plosgenetics.org/article/info:doi/10.1371/journal.pgen.1003740

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