Researchers look into animal genomes in search of causes of human diseases

Precision medicine still struggles with a basic problem: understanding how genomic context can amplify or mute the effects of isolated mutations. And so, precision medicine is more than willing to take lessons from other disciplines, namely, comparative genomics and evolutionary biology. These disciplines may help explain why mutations that are frequently pathogenic in the human genome are harmless in other genomes. For example, they may identify genomic features that compensate for potentially disease-causing mutations.

Making sense of genomic context was at the heart of a recent study conducted by researchers from Duke University and Brigham and Women's Hospital. In this study, the researchers compared thousands of human disease-causing mutations with the analogous sequences of some 100 animal species.

Focusing on missense variants, the researchers found that an appreciable fraction of disease-causing alleles are fixed in the genomes of other species, suggesting a role for genomic context. Although generally in line with earlier observations, the scale of the findings motivated geneticists to find the explanation for this apparent mystery.

"We found many examples in which an entire species should have a serious genetic ailment, but instead were healthy," said Duke's Nicholas Katsanis, Ph.D. "So, if we can understand how animals escape illness from such severe genetic mutations, we might have a way to make humans better. What we considered is that for many mutations, there must be a buffering mechanism—another mutation that protects the animal from the detrimental effects of the disease-causing mutation."

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis. Read full, original post: Animals' Genomic Buffers May Help Humans