Mutational 'hot spot' leads to adaptation in high-altitude birds

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Jay Storz. Credit: Craig Chandler/University Communications

New research led by the University of Nebraska-Lincoln and the University of New Mexico has pinpointed changes in a single, mutation-prone gene site that ultimately allow wrens to breathe easy in the rarefied air of the Andes.

Published Oct. 12 in the journal *Proceedings of the National Academy of Sciences*, the findings also suggest that a gene site's rate of mutation may influence whether it contributes to adaptive evolutionary changes.

The authors concluded that when many mutations can produce the same favorable change in a trait, natural selection often seizes upon those that occur at an especially high rate—an evolutionary equivalent of "first come, first served."

The team's study of Andean wrens examined evolutionary changes in the functional properties of hemoglobin, the protein that transports oxygen from the lungs to body tissue. Oxygen concentration falls as altitude rises, making the protein's ability to capture oxygen molecules especially critical among wrens and other animals that live at higher elevations.

"We know from previous research that there are many possible mutations that can produce an increase in hemoglobin's affinity for oxygen," said UNL co-author Jay Storz, a Susan J. Rosowski Professor of Biological Sciences. "However, the particular mutations that actually contribute to evolutionary changes in protein function may represent a biased, nonrandom subset of all possible mutations capable of producing the change.

"Our results suggest that variation in mutation rate may be an important source of such bias."

Storz and his UNL colleagues came to the conclusion after analyzing blood samples from 140 house wren specimens that ornithologist Christopher Witt and fellow UNM researchers collected during an expedition to the Andes.

The UNL researchers isolated hemoglobin proteins from the blood samples, comparing those that had the specified mutation against those that did not. The team also engineered synthetic hemoglobin that featured only the mutation of interest. Storz and his colleagues found that synthetic proteins with the engineered mutation captured oxygen roughly 1 1/2 times better than those without it.

The researchers also measured the frequency of the affinity-enhancing mutation in highland versus lowland wrens, discovering that the mutation predominated only in the high-altitude population.

To test whether the altitudinal difference in mutation frequency was caused by natural selection, the team then examined patterns of variation at DNA sites scattered throughout the species' genome.

"The genomic analysis revealed that the level of altitudinal differentiation at that single mutational 'hot spot' is much larger than what we see across the genome as a whole," Storz said. "That pattern suggests that the site-specific differentiation was caused by natural selection that favored different protein variants in different elevation zones."

More information: Contribution of a mutational hot spot to hemoglobin adaptation in high-altitude Andean house wrens, www.pnas.org/cgi/doi/10.1073/pnas.1507300112

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