Foot feathering in domesticated breeds of pigeons and chickens use same gene regulatory networks

April 29 2020

Not only are the same genes involved in foot feathering phenotypes in pigeons

and chickens, but the nature of the mutations also very much the same. The study reveals a remarkable convergence in the evolution of the feathered leg phenotype in domestic chickens and domestic pigeons as this phenotype is caused by non-coding mutations upstream of the same two genes. Credit: USDA

Poultry geneticists have long studied the inheritance of a prized fancy chicken breeding trait; feathered legs.

Lead researcher Leif Andersson and colleagues at Texas A&M University, have investigated the genetic basis of foot feathering. The trait, known scientifically as ptilopody, can be observed in domesticated and wild avian species and is characterized by the partial or complete development of feathers on the skin of the ankles and feet.

"Foot feathering is an interesting case since, although it is a very recognizable trait that can be very easily selected to fixation in breeds, it is in fact not a monogenic trait, caused by a single gene," said Andersson.

In previous studies, two independent dominant alleles, denoted Pti-1 and Pti-2, were found to cause feathered legs in domestic chicken. Breeds with more sparsely feathered legs were supposed to be homozygous mutant (two copies) at the Pti-1 locus but wild-type (pti-2/pti-2) at the second locus. Breeds with more heavily feathered legs like Sultan and Cochin were thought to carry the dominant mutations at both Pti-1 and Pti-2.

In addition, other studies had demonstrated that regulatory mutations in the genes Tbx5 and Pitx1 contribute to foot feathering in domesticated breeds of pigeon.

Andersson and his colleagues wanted to find out which genes and specific mutations correspond to the previously described Pti-1 and Pti-2 loci contributing to foot feathering in chickens—-and if these are the same genes as previously identified in pigeon.

In evolution, parallel evolution is known as the independent development of similar phenotypic traits in separate but related lineages. In other words, when faced to adapt, nature can go to the same genetic toolbox to come up with new solutions.

Now, in the advanced online edition of the journal *Molecular Biology and Evolution*, Andersson and co-authors report that indeed, regulatory mutations in the same genes are responsible for foot feathering in domesticated chickens.

To perform the study and identify the casual mutation of the feathered leg trait in chickens, a three-generation mapping population was established by crossing Langshan (feathered leg) and Houdan (clean leg) chickens, followed by SNP analyses of pooled samples, linkage mapping, whole genome sequencing (WGS), and diagnostic testing of a comprehensive collection of 167 chicken populations with or without feathered legs.

The first predicted causal mutation is a single base change located 25 kb upstream of the gene for the forelimb-specific transcription factor TBX5 on chromosome 15. The second is a 17.7 kb deletion located about 200 kb upstream of the gene for the hindlimb-specific transcription factor PITX1 on chromosome 13. These mutations are predicted to activate TBX5 and repress PITX1 expression, respectively. Furthermore, the PITX1 causal variants are large overlapping deletions, 17.7 kb in chicken and 44 kb in pigeons.

"The identification of causal mutations in non-coding parts of vertebrate

genomes is challenging, due to the difficulty in deducing or experimentally proving functional significance," said Andersson. "Here we have presented strong genetic evidence for causality for two non-coding mutations affecting the feathered leg phenotype in domestic chicken using a very large data-set from chicken with or without feathered legs."

Thus, not only are the same genes involved in foot feathering phenotypes in pigeons and chickens, but the nature of the mutations also very much the same, at least in the case of Pitx1.

The study reveals a remarkable convergence in the evolution of the feathered leg phenotype in domestic chickens and domestic pigeons as this phenotype is caused by non-coding mutations upstream of the same two genes.

"These two cases add to a growing list of regulatory mutations controlling phenotypic traits in domestic animals by altering the expression of important transcription factors," said Andersson. "In fact, the genetic basis of leg feathering is a parallel example to the genetic basis for variation in comb morphology in chickens. Three comb phenotypes Pea-comb, Rose-comb and Duplex-comb, are all structural variants leading to ectopic expression of respectively the SOX5, MNR2, and EOMES transcription factors during comb development."

The manuscript provides strong evidences that foot feathering in chickens has a genetic determinism highly similar to the very same trait in pigeon and confirms that ptilopody has evolved through by running on separate but parallel tracks in chicken and pigeon.

"There is a remarkable convergence as regards the evolution of the feathered leg phenotype in domestic chicken and in domestic pigeons, said Andersson. "The present study illustrates that novel phenotypic

traits in domestic animals are often caused by the same mutations across divergent populations because favored mutations have been spread from population to population due to strong phenotypic selection The extensive collection of whole genome sequence data, which is publicly available, provides a powerful resource to identify genomic regions and corresponding causal mutations associated with these phenotypes as successfully accomplished in this study."

"Furthermore, feathered legs occur in many other bird species, for instance in most owls but not in all. If you would like to study the genetic basis for variation in leg feathering within a species or between closely related species, Tbx5 and Pitx1 are the obvious candidate genes to start with," said Leif Andersson.

More information: J Li et al, Mutations upstream of the TBX5 and PITX1 transcription factor genes are associated with feathered legs in the domestic chicken, *Molecular Biology and Evolution* (2020). DOI: 10.1093/molbev/msaa093

Provided by Oxford University Press