

# Case Study 23

# Parallel evolution of foot feathering in birds is mirrored by parallel evolution of genes

**Leading IMAGE partner: WU**Other IMAGE partners involved: FLI

Other non-IMAGE partners and actors involved: Fancy breeders provided samples

IMAGE WP and Task: WP4, Task 4.3

## **Objectives**

Understanding the genetic basis of similar phenotypes shared between lineages is a long-lasting research interest. Even though animal evolution offers many examples of parallelism, for many phenotypes little is known about the underlying genes and mutations. The aim of this case study was to investigate the genetic basis and parallel genetic origin of ptilopody (*Pti*), or foot feathering, in chicken.

## Material and methods

Whole-genome sequencing data from 91 traditional Dutch chicken breeds and 68 traditional German chicken breeds were used. Transcriptomic data from four traditional Dutch chicken breeds were also generated on a later stage to validate our results. Potential genomic regions associated with the phenotype were identified through a genome-wide association analysis (GWAS). Comparative genomic approaches were used to investigate the level of conservation and evolution of the genomic regions associated with the phenotype. Differentially expressed genes were identified using DESeq2.

#### Results/output

Two genomic regions were identified to be associated with ptilopody. At one of the loci, we identified a 17-kb deletion affecting *PITX1* expression, a gene known to encode transcript regulators of hindlimb identity and development. Similarly to pigeon, at the second loci we observed ectopic expression of *TBX5*, a gene involved in forelimb identity and a key determinant of foot feather development. We also observed that the trait evolved only once as foot-feathered birds share the same haplotype upstream *TBX5*. Our findings indicate that in chicken and pigeon ptilopody is determined by the same set of genes that affects similar molecular pathways. Our study confirms that ptilopody has evolved through parallel evolution since the rise of *de novo* mutations in the ancient history of bird evolution.





#### **More information**

Chiara Bortoluzzi, Hendrik-Jan Megens, Mirte Bosse, Martijn F L Derks, Bert Dibbits, Kimberly Laport, Steffen Weigend, Martien A M Groenen, Richard P M A Crooijmans, 2020. Parallel Genetic Origin of Foot Feathering in Birds, *Molecular Biology and Evolution*, msaa092, https://doi.org/10.1093/molbev/msaa092.

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