

Detection and characterization of deleterious variants in traditional chicken breeds

Chiara Bortoluzzi¹, Martijn Derks¹, Steffen Weigend², Martien AM Groenen¹, Hendrik-Jan Megens¹

¹Wageningen University & Research (Netherlands), ²Institute of Farm Animal Genetics, Neustadt- Mariensee (Germany)

Deleterious variants are constantly generated by mutation, but are not always purged from the population. Understanding the extent and the reasons why deleterious variants persist in populations are of interest for conservation purposes, because these variants reduce reproductive fitness and genetic diversity when homozygous, which becomes especially relevant when populations decline.

Traditional chicken breeds offer a powerful model to address the role of demographic discontinuities and selection on deleterious variation, because of their intense artificial selection, small effective population size, and substantial genetic variation. Here, we describe and characterize the genome-wide distribution of low frequency deleterious and loss-of-function (LoF) variants in protein-coding genes of 76 whole-genome sequences from traditional breeds that represent different management, demographic, and selection histories, and conservation status. On average, we observed 4774 missense variants, of which 8.42% were deleterious and 70% tolerated. We identified 1203 stop-gain, 1648 frame-shift, 687 start-lost, and 715 stop-lost variants. A Gene ontology (GO) overrepresentation test showed that genes with deleterious and LoF variants were involved in the regulation of development processes, reproductive system development, ovarian follicle development, and oviposition. Regions of the genome under selection enriched for missense and LoF variants were involved in body weight and skeletal integrity.

We show that deleterious and LoF variants affect gene products involved in vital development and reproductive functions. Our findings confirm the importance of next-generation sequencing information in the design of breeding schemes and conservation programs.
