

CASE STUDY 5

Persistence of small populations depends on the type of population contraction

Leading IMAGE partner: WUR

Other IMAGE partners involved: None

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

IMAGE WP and Task: WP4, Task 4.3

Objectives

Predictions about the consequences of a small population size on genetic and deleterious variation are fundamental to population genetics. As small populations are more affected by genetic drift, purifying selection acting against deleterious alleles is predicted to be less efficient, therefore increasing the risk of inbreeding depression. However, the extent to which small populations are subjected to genetic drift depends on the nature and time frame in which the bottleneck occurs. The aim of the study was to directly examine the consequences of two types of bottlenecks for deleterious variation: the severe domestication bottleneck and the recent population decline accompanying breed formation.

Material and methods

Whole-genome sequencing data (WGS) of 97 individuals from 37 traditional Dutch chicken breeds were used. Genetic diversity was measured along the genome as level of heterozygosity. Regions of reduced diversity (or runs of homozygosity - ROHs) were used as a measure of inbreeding. Harmful mutations were identified with the Variant Effect Predictor (VEP) program and analyzed in relation to the runs of homozygosity

Results/output

The size of a population is an important factor in determining the level of genetic variability and the effectiveness of selection to remove harmful mutations. In particular, populations that since the ancient domestication bottleneck have been kept small in size seem to have eliminated harmful mutations of very strong effect. On the contrary, in recently bottlenecked populations the accumulation of weakly harmful mutations is mainly driven by stochastic demographic and genetic events, which overrule selection against them. In recently bottleneck populations harmful mutations were also mostly found in long regions of the genome with reduced variation (runs of homozygosity), suggesting that this clustering is likely to occur due to stochastic effects and increased chances of mating between related individuals (i.e. inbreeding).

Even though (wild and domesticated) small populations are of conservation concern, conservation or breeding programmes are often not in place. Therefore, the establishment of such programmes that combine genomic information on harmful variation with inbreeding avoidance is the first step to assure the long-term survival of small populations, while enhancing their genetic diversity.

More information

Results of this case study were presented at the EAAP 2019 conference in Ghent and published in *Evolutionary Applications*, see <https://onlinelibrary.wiley.com/doi/epdf/10.1111/eva.1287>.

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