

# A comprehensive detection and characterization of deleterious variants in traditional chicken breeds

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## Background

Deleterious variants are constantly generated by mutation (Fig 1), but are not always purged from the population.

Understanding the extent and the nature of deleterious variation in small populations is of interest for conservation purposes, because these variants **reduce reproductive fitness** (Fig 2) and **genetic diversity** when homozygous.

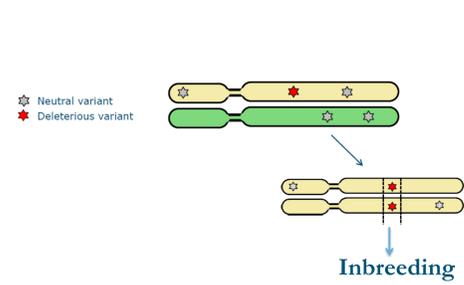


Figure 1. An example of deleterious variants

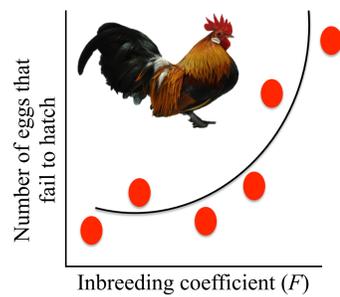


Figure 2. Reduction in reproductive fitness due to the accumulation of deleterious variants as result of inbreeding

Traditional chicken breeds offer a powerful model to address the role of demographic discontinuities and selection on deleterious variants

## Results

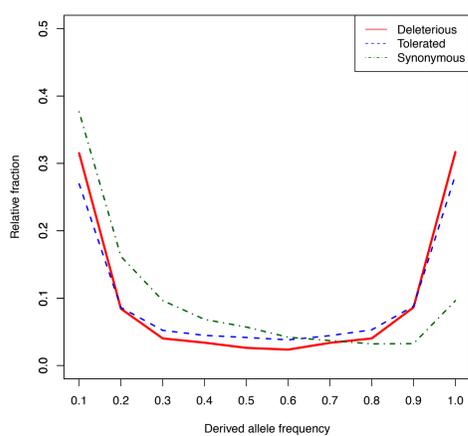


Figure 4A. Derived allele frequency of synonymous and deleterious variants

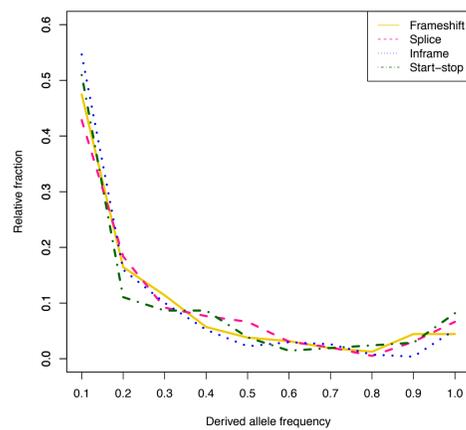


Figure 4B. Derived allele frequency of loss of function (LoF) variants

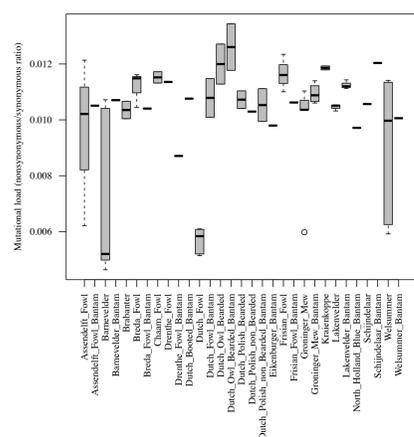


Figure 5. Mutational load of homozygous derived alleles

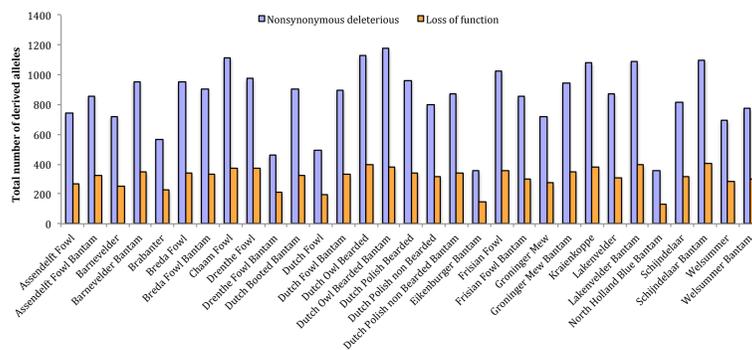


Figure 6. Total number of derived (heterozygous and homozygous derived) alleles for deleterious and loss of function variants

We developed a **comprehensive catalogue of putatively deleterious variants** for traditional Dutch chicken breeds. Such genomic catalogues can be used to enhance the conservation status of traditional breeds by lowering the frequency of undesirable variant in the population.

## Materials & Methods

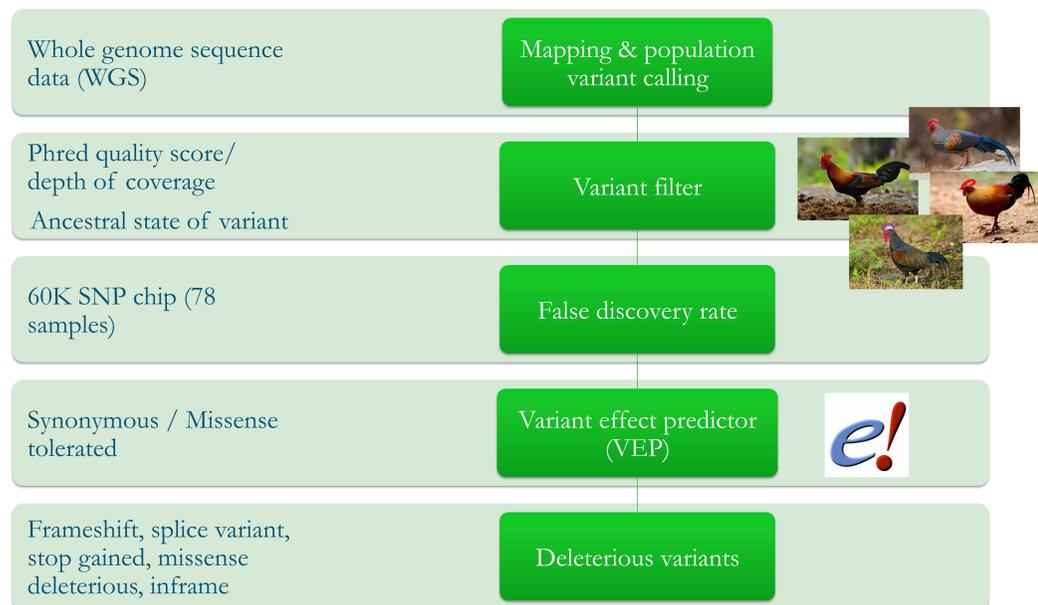


Figure 3. Pipeline overview used to detect deleterious variants using whole-genome sequence data

## Discussion

The predicted deleterious variants are **subject to purifying selection**, as confirmed by the skewed distribution towards a higher proportion of low-high derived allele frequency alleles (Fig. 4A, Fig. 4B).

Mutational load considerably varies across traditional breeds as a result of their different demographic and management history (Fig. 5).

Genotypes containing putatively derived alleles are lowest for deleterious and LoF variants (Fig. 6) relative to synonymous and missense tolerated (Fig. 7). This further suggests that purifying selection effectively removes deleterious variation even in population characterised by small effective population size.

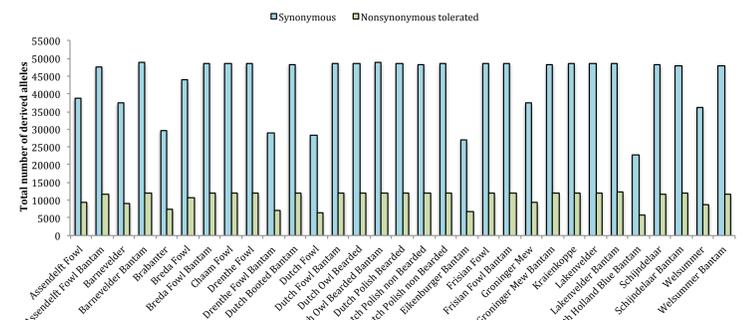


Figure 7. Total number of derived (heterozygous and homozygous derived) alleles for synonymous and missense tolerated variants