Determinants of genetic load in natural populations of guppy (*Poecilia reticulata*)

PhD thesis-abstract

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The term genetic load describes deleterious variation in a population which causes fitness decrease in the present or potentially might cause such decrease in the future. Demographic process can lead to accumulation of such variants, when a bottleneck takes place and population size is reduced. This happens because in small populations forces of drift dominate the natural selection, so slightly deleterious alleles can increase in frequency. Bottlenecked populations are also characterized by increased homozygosity due to inbreeding, inevitable in small populations. Inbreeding might also have a positive effect on population fitness. If highly deleterious mutations are recessive, it can expose them to the selection in homozygotes, effectively decreasing their frequency. A relative role of relaxed selection and purging of recessive variants is heavily discussed, particularly, its role in both invasive and vulnerable species. Among species listed as endangered, majority has small census size and low genetic diversity. Most of them share common history of bottlenecks, could have led to load accumulation, but also to purging. Another important issue regarding genetic load is its role in biological invasions. The beginning of species invasion is marked with small population size, so it might accumulate deleterious variation on the spread axis. However, it may also purge harmful variants, increasing its invasive potential.

In the first chapter, I estimated per site, per generation mutation rate in guppies. Since new generation sequencing is error prone, candidate *de novo* mutations were then filtered based on a set of criteria to distinguish positives from negatives. As such filtering criteria are subjective, I decided to separately test alternative approach, based on machine learning. After comparing effectiveness of the two approaches, I found that both methods estimated similar mutation rate, but their false negative and false discovery rates differed substantially. The machine learning approach found more true positive *de novo* mutations, but was more effort-consuming. Finally, I found that guppy mutation rate is low – 2.9×10^{-9}, which is in compliance to other Teleostei fish estimates.

The second chapter of my thesis explores genetic load in 14 wild guppy populations from Trinidad and Tobago. I particularly focused on river Turure, where expansion is taking place. It is a result of an introduction of guppies into a guppy-free location in the upstream, which was followed by a downstream propagation and admixture with native individuals in the lower parts. The expanding population was successful and replaced original inhabitants of downstream Turure. To find whether the invasion facilitated purging of genetic load during initial bottleneck, or on contrary, was followed by accumulation of genetic load, I studied three sites along the river. I also explored load of populations differing in size, contrasting large populations with small populations as well as upstream and downstream rivers locations. Finally, I looked for relationship between N_e and load across populations. Downstream and middle Turure populations were less loaded than the population from upstream location, therefore I found no proof of genetic load accumulation along the spread axis. This observation could be explained by admixture between migrants and locals, which alleviated effects of initial bottleneck. The upstream population had the highest load among all Turure locations and similar amount of high effect variants, so there was no indication of purging during the initial stage of invasion. When it comes to populations from other rivers, I observed that small populations and big populations differ in terms of load, with the former having higher burden. Similar situation was found in more loaded post-bottleneck upstream populations and less loaded downstream populations within the same rivers. In this case, however, the difference in effective population size failed to explain the difference in load. Such size-dependent conclusion could not have been made generally as well, since no relationship between N_e and load was found across majority of populations. This however, could have been due to large N_e of studied populations.

Overall, this thesis indicates that while genetic load tends to accumulate in bottlenecked populations, gene flow can significantly relieve the burden and loosen association of the load with N_e. It also demonstrates that guppies, like other Teleostei, are characterize by low mutation rate, what can significantly affect amount of variation present in this species.

Keywords: mutation rate, guppy, genetic load, expansion