



Role of Phenotypically-Informative SNP Markers in Conservation Biology

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Anthropogenically induced environmental change is causing populations to go extinct at unprecedented rates. This has created a triage situation where the preservation of genetically distinct populations could be prioritized if methods could be developed to quickly identify them. Traditional “common garden” methods of identifying traits showing genetic differences among populations are often too slow to be useful. I will argue that modern genomics is enabling discovery of DNA markers that are phenotype informative for skin pigmentation patterns, coat color, eye color and body size that could be used as an index of genetic distinctness and therefore conservation priority. I believe that the potential role of phenotypically informative SNPs has not previously been recognized because of the paradigm that local adaptation mostly involves quantitative traits determined by many genetic loci (QTL) of small effect.

The development of high density SNP (single nucleotide polymorphism) chips for humans and increasing interest in molecular forensics has facilitated the discovery of the phenotypically informative mutations for skin colour, eye colour, and hair colour [1]. A number of SNPs in two highly conserved genes: the melanocortin 1 receptor (MC1R) gene and the agouti signaling protein (ASIP) gene have been shown to affect not just human pigmentation but also that of other vertebrates such as deer mice and fishes [2]. Two SNPs in the MC1R gene cause a blue variant of the arctic fox whose coat colour does not become white in winter [3]. The blue variant may be adaptive in Iceland, Svalbard and western Greenland where some individuals have adapted to a coastal life-style [4] and where coat colour could be noninvasively detected by genotyping of fox faecal pellets.

High density genotyping and genome wide association analysis (GWAS) makes it possible to find SNPs that are physically linked to ecologically important traits controlled by major loci [5] that are under selection for different optima in different environments. For example, juvenile salmon from gravel-bottomed streams have dark vertical bars on their sides while those from mud-bottomed streams have faint or no marks; these skin patterns help camouflage parr from bird predators against their natal streams [6]. The number and the contrast of parr marks seem to be controlled by different sets of major QTL [7] as well as limited phenotypic plasticity. Atlantic salmon SNP chips containing 5000 to 200,000 polymorphic SNPs can be used in

wild populations [8] increasing the likelihood that genome regions responsible for adaptive differences among rivers will be tightly linked to specific SNP markers (Boulding, unpublished data). This may eventually allow us to use SNP genotypes to predict adaptation trait values of extirpated Atlantic salmon populations from archived scale samples as is now done for ancient human skeletal remains [9].

Detection of local adaptation with phenotypically informative SNPs in endangered, non-model organisms is becoming more feasible as high-throughput SNP genotyping becomes possible for more species and as efficient GWAS methods that use linkage disequilibrium among small populations become more sophisticated. Some ecologically relevant traits are determined solely by small QTL and therefore the discovery of phenotypically informative useful SNPs is unlikely. This could create a possible trait bias in any index of genetic distinctness. Phenotypically informative SNPs are more likely to be discovered for traits determined by major QTL. Major QTL are more likely to persist in when selection on the trait is diversifying rather than directional. A promising area for future research is to explore the extent to which phenotypically informative SNPs can be used as an index of genetic distinctness and therefore conservation priority. Phenotypically informative SNPs should be especially useful in field situations where the phenotype is not easy to observe.

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