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Genomics and the challenging translation into conservation practice

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The global loss of biodiversity continues at an alarming rate. Genomic approaches have been suggested as a promising tool for conservation practice as scaling up to genome-wide data can improve traditional conservation genetic inferences and provide qualitatively novel insights. However, the generation of genomic data and subsequent analyses and interpretations remain challenging and largely confined to academic research in ecology and evolution. This generates a gap between basic research and applicable solutions for conservation managers faced with multifaceted problems. Before the real-world conservation potential of genomic research can be realized, we suggest that current infrastructures need to be modified, methods must mature, analytical pipelines need to be developed, and successful case studies must be disseminated to practitioners.

Conservation biology and genomics

Like most of the life sciences, conservation biology is being confronted with the challenge of how to integrate the collection and analysis of large-scale genomic data into its toolbox. Conservation biologists pull from a wide array of disciplines in an effort to preserve biodiversity and ecosystem services [1] and genetic data have helped in this regard by, for example, detecting population substructure, measuring genetic connectivity, and identifying potential risks associated with demographic change and inbreeding [2]. Traditionally, conservation genetics (see Glossary) has relied on a handful of molecular markers ranging from a few allozymes to dozens of microsatellites [3]. However, for close to a decade [4] genomics – broadly defined as high-throughput sampling of nucleic acids [5] – has been touted as an important advancement in the field, a panacea of sorts for the unresolved conservation problems typically addressed with genetic data [6,7]. This transition has led to much promise but also hyperbole, where concrete empirical examples of genomic data having a conservation impact remain rare.

Under the premise that assisting conservation of the world's biota is its ultimate purpose, the emerging field of conservation genomics must openly and pragmatically discuss its potential contribution toward this goal. While there are prominent examples where genetic approaches have made inroads influencing conservation efforts (e.g., Florida panther augmentation [8,9]) and wildlife enforcement (i.e., detecting illegal harvesting [10]), it is not immediately clear that the conservation community and society more broadly have embraced genomics as a useful tool for conservation. Maintaining genetic diversity has largely been an afterthought when it comes to national biodiversity policies [11,12] and attempts to identify areas that might prove to be essential for conserving biological diversity rarely mention genomics (e.g., [13,14]). An obvious reason for this disconnect is that many of the pressing conservation issues (e.g., [15,16]) simply do not need genomics but instead need political will.

The traditional use of genetic data in conservation biology has been historically demarcated into two interrelated areas [3]: (i) understanding how evolutionary processes such as genetic drift, selection, and migration shape genetic and phenotypic variation of natural populations and determine population structure; and (ii) more specifically, describing the effects of low effective population size on genetic variation and population viability. Nested within these are more general conservation issues such as resolving taxonomic uncertainties, preserving local adaptation, and offsetting inbreeding depression (Table 1). Whether genome-scale data can improve inferences within these two areas and better inform conservation initiatives remains up for debate. Furthermore, there are a plethora of uncertainties that practitioners need to be aware of, and considerable obstacles that need to be overcome, before genomics can make the transition to applied conservation science. Many of the qualitatively novel aspects of genomic analyses, which include monitoring of epigenetic markers [17], environmental DNA approaches to assay species communities [18], and transcriptome assays [19], are still at an exploratory stage and are far from seeing use in real-world conservation issues.

Here we focus first on how traditional applications of genetics in conservation can benefit from scaling up to genome-wide data. In particular, we highlight two key areas that have received attention in the literature:

Glossary

Adaptive locus: a region of the genome under selection that encodes a phenotype (or is closely linked to a causative locus) with fitness consequences in a particular environment.
Annotation: the process of delineating and assigning function to genetic sequences.
Background selection: the loss of genetic diversity at neutrally evolving sites that are linked to sites under purifying selection.
Candidate genes: genes putatively underlying variation in a certain phenotype.
Coalescent theory: a retrospective population genetics framework that traces genetic variants of a locus to the most recent common ancestor. Used to infer demographic parameters of population histories.
Conservation genetics: uses genetic markers to help conserve biodiversity and manage species and populations. Traditional genetic markers include allozymes, microsatellites, and targeted gene sequences.
Conservation genomics: uses genome-wide information to help conserve biodiversity and manage species and populations. Genomic data is derived from high-throughput sequencing technology. Relevant examples are whole genome resequencing and targeted approaches like exome sequencing, GBS, SNP genotyping, and transcriptome sequencing.
Effective population size: a population genetics convention describing the number of breeding individuals in an ideal population that would lose genetic variation at the same rate as the observed population.
Environmental DNA: DNA found in environmental samples (e.g., water, soil) that can be used in genetic or genomic analysis. This contrasts with traditional approaches that target a specific organism or tissue.
Genetic drift: the loss of genetic variants due to random sampling from one generation to the next.
Genome assembly: the process of ordering and orienting sequencing into a contiguous consensus sequence of the genome.
Genotyping by sequencing (GBS): the sequencing of a repeatable subset of the genome seeded by restriction enzyme recognition sites. Restriction site-associated DNA sequencing (RAD-seq) is another commonly used term.
Haplotypes: particular combinations of alleles at collinear positions along a stretch of DNA.
Inbreeding: the increase of genomic segments in identity by descent due to mating between closely related individuals. Results in an increase in homozygosity, potentially revealing detrimental recessive alleles with negative fitness consequences.
Linkage disequilibrium: the non-random association of alleles at two or more loci.
Orthology: homologous DNA sequence descended from a shared common ancestor.
Outlier locus: a region of the genome that, based on user-defined criteria (often extreme population differentiation), deviates from the rest of the entire genome.
Recombination: the process of genetic exchange between homologous chromosomes, often resulting in a new combination of alleles.
Transcriptome: the set of all RNA molecules transcribed from a DNA template.