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DNA-based studies and genetic diversity indicator assessments are complementary approaches to conserving evolutionary potential

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Abstract

Genetic diversity is essential for maintaining healthy populations and ecosystems. Several approaches have recently been developed to evaluate population genetic trends without necessarily collecting new genetic data. Such “genetic diversity indicators” enable rapid, large-scale evaluation across dozens to thousands of species. Empirical genetic studies, when available, provide detailed information that is important for management, such as estimates of gene flow, inbreeding, genetic erosion and adaptation. In this article, we argue that the development and advancement of genetic diversity indicators is a complementary approach to genetic studies in conservation biology, but not a substitute. Genetic diversity indicators and empirical genetic data can provide different information for conserving genetic diversity. Genetic diversity indicators enable affordable tracking, reporting, prioritization and communication, although, being proxies, do not provide comprehensive evaluation of the genetic status of a species. Conversely, genetic methods offer detailed analysis of the genetic status of a given species or population, although they remain challenging to implement for most species globally, given current capacity and resourcing. We conclude that indicators and genetic studies are both important for genetic conservation actions and recommend they be used in combination for conserving and monitoring genetic diversity.

Keywords Biodiversity monitoring · Genomics · Population genetics · Prioritisation

Introduction

Several approaches have recently been developed and used to efficiently and rapidly evaluate the genetic status and trends of populations and species, in the absence of collecting new genetic data (in this article we use “genetic data” and “genetic studies” to mean molecular techniques to examine the DNA or similar studies of inherited variation, e.g. DNA sequencing). Building on decades of prior work (Wright 1943; Frankham 1995; Manel et al. 2003; Hamann et al. 2005; Hanson et al. 2017), these approaches leverage population genetic theory and proxy-based geographic and

demographic data to approximate genetic status and trends. For example, two indicators were developed and adopted as part of the United Nations Convention on Biological Diversity (CBD) Global Biodiversity Framework (GBF): 1) the proportion of populations large enough to maintain genetic diversity (i.e., effective population size, N_e , > 500), and 2) the proportion of populations maintained (Laikre et al. 2020; Hoban et al. 2020, 2023a). These indicators are both valuable because they reflect different aspects of genetic diversity, and one or both may be derived from existing ecological monitoring data, if available (Mastretta-Yanes et al. 2024a). Other simple and policy-relevant genetic indicators have been developed, such as a multi-dimensional genetic health index (Kriesner et al. 2020), genetic scorecards for

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high-profile species that document possible genetic threats and ongoing actions (Hollingsworth et al. 2020; O'Brien et al. 2022), and indicators of the sufficiency of protection (i.e., safeguarding) of genetic diversity across species' ranges (Khoury et al. 2019, 2020). The latter two are also indicators under the GBF. All of these indicators can use existing non-genetic data to varying degrees, along with summaries of previous genetic datasets when available, to assess the genetic status, trends, and/or effectiveness of conservation actions for species and populations, without the collection of new genetic (e.g. DNA-based) information. The use of indicators therefore allows rapid evaluation and comparison among dozens to thousands of species using simple, repeatable and accessible metrics with available data.

Here, we aim to emphasize that the development and advancement of such indicators complement genetic studies, that generate and analyse genetic information (including "genomic" approaches e.g. high throughput DNA sequence data; Fig. 1), and do not reduce the pressing need for studies that collect genetic data. When genetic data are feasible to obtain, they can be informative for specific cases, while also contributing to the growing evidence-base that underlies

pragmatic indicators. Genetic diversity indicators and genetic studies each provide different types of information for different uses, and *both are vital for informing the conservation of genetic diversity*. Indicators are especially useful for rapid, large-scale assessments when genetic data are not comprehensively available, and support policymakers and other stakeholders in prioritizing management among many species and populations using a common metric. In turn, these data can help direct resources towards species or populations in greatest need of more in-depth investigation, and support communicating to the public about genetic concepts and the urgency of genetic conservation (see Table 1).

Genetic diversity indicators

The genetic indicators included in the CBD GBF were originally designed to enable tracking of genetic status across many species and monitoring change over time, especially for the many species without genetic data, within the context of policy and management. One of the weaknesses of the previous CBD Aichi Biodiversity Targets was a lack of

Fig. 1 Genetic studies (typically using DNA data) and genetic indicator assessments (based on new and existing diverse datasets, such as ecological surveys) provide complementary and distinct information for guiding the genetic management of biodiversity

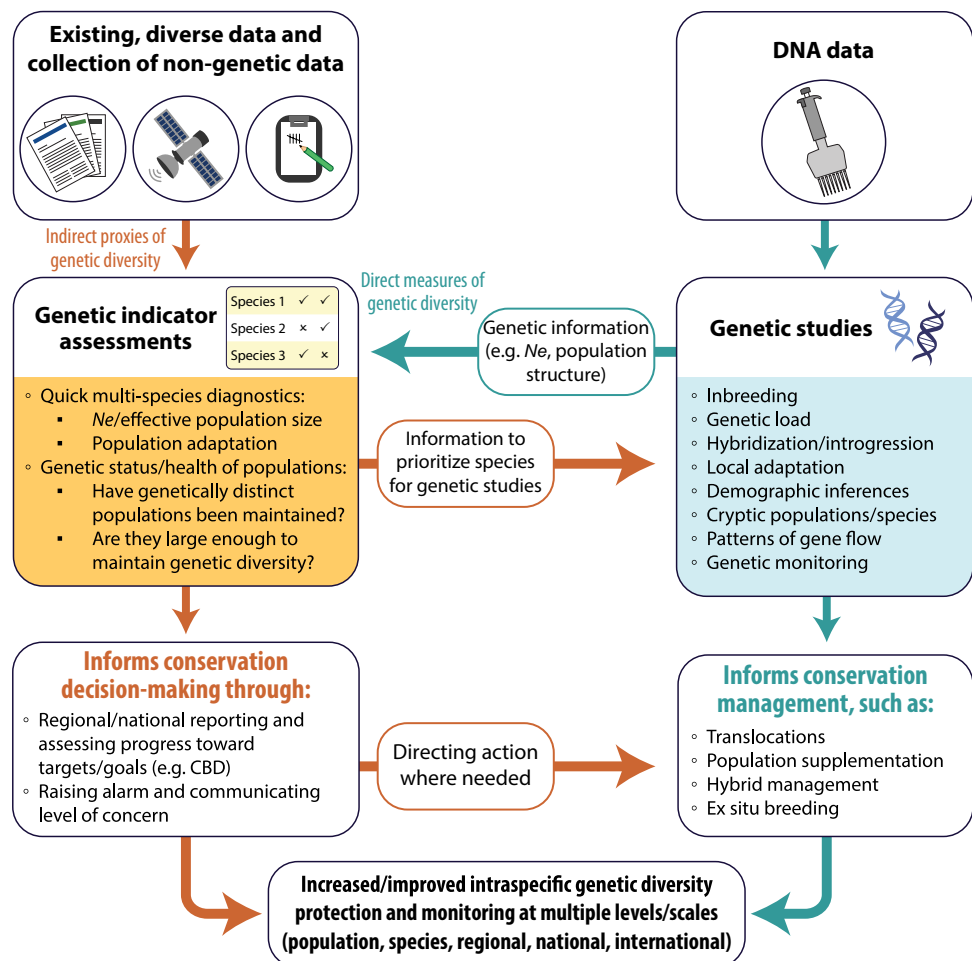


Table 1 Multiple assessments of genetic diversity indicators across moderate to large numbers of species have been conducted and have generated policy or management guidance, quickly, by using existing data. Genetic data and proxies of genetic diversity were integrated to varying degrees in each of these analyses

Number of species assessed	Conclusions and use for policy and management	Citation
909	Many species retain a majority of their populations, but most populations are below the threshold of $N_e > 500$ and may be in danger of genetic diversity loss. The indicator highlights populations and species in need of active intervention	Mastretta-Yanes, da Silva, et al. (2024a)
7,336 and 4,470	In a first-pass evaluation of genetic indicators in Sweden, 60% of 7,336 species potentially have populations with census size $N_c > 5,000$, while 91% of 4,470 species have lost some subpopulations or distribution during the last 100 years	Thurfjell et al. (2022)
594	Crop wild relative genetic diversity has not been well conserved ex situ or in situ, and most species require urgent action. The indicator provides a quantitative measure of progress on this action (geographic or ecological range conserved)	Khoury et al. (2020)
26	Assessment of several criteria show that 14 species are likely at negligible genetic risk, eight species at moderate risk (with effective mitigation in place for five of these), and four species are likely at risk of severe genetic problems	Hollingsworth et al. (2020)
1,148	Approximately 15–20% of assessed species are “likely to benefit from... genetic rescue or targeted gene flow/genetic augmentation. Over half... would benefit from re-establishing new populations”	Kriesner et al. (2020)
1,321	Documents that genetic diversity (GD) is reduced in threatened species and proposes a straightforward framework to rank species based on publicly available GD data and time to expected loss of GD due to small populations	Willoughby et al. (2015)
83	Mean individual zygosity (i.e., heterozygosity and autozygosity) estimated from publicly available resequencing data can be used as a key criterion in a novel analytical framework to help rank species by conservation category	Jeon et al. (2024)

quantitative, reliable indicators (Butchart et al. 2016; Xu et al. 2021), and so these were required in the more-recent CBD GBF. Although genetic diversity has been overlooked in conservation policy in the past (Laikre et al. 2010), genetic diversity indicators enabled this important component of biodiversity to be included in the GBF (Hoban et al. 2023a; Carroll et al. 2023). Genetic indicators have some notable limitations, such as focusing primarily on only two aspects of genetic health (amount of genetic drift and numbers of putatively locally adapted populations), utilizing thresholds (such as $N_e > 500$) rather than continuous metrics, and relying on assumptions such as population-level adaptation and a universal estimate of the N_e/N_c ratio. However, these indicators enable biodiversity managers to quantify likely genetic change for the thousands of species for which detailed genetic studies are not currently available or likely to be so in the near future, and manage many populations that may need intervention (Hoban et al. 2023b, 2024; Mastretta-Yanes et al. 2024a). Indicators can thus enable the following key outcomes: tracking change over hundreds of species and across the world; reporting such change under policy frameworks including the CBD and regional and national frameworks where hundreds or thousands of species (and often their distinct populations) are protected and assessed (e.g., U.S. Endangered Species Act, Canadian Species at Risk Act, European Habitats Directive, European Nature Restoration Law, Mexican SEMARNAT NOM-059, South African National Environmental Management:

Biodiversity Act); prioritizing—based on a common metric—species that have the most urgent conservation needs to remedy genetic diversity loss; and facilitating communication on genetic issues to non-geneticists, the public and policymakers. Further research on genetic diversity indicators, including complementary studies with genetic data, will help improve indicators into the future.

Genetic data, notably from DNA-based studies

Genetic data have been used for decades to study the genetic “health” of species in detail, and the processes impacting the conservation of genetic diversity (Frankham 2010). Genetic data can reveal diverse and detailed information, such as understanding hybridization, population distinctions or barriers to gene flow, ancient population size and size changes (and thus providing context for current size), assessing inbreeding at the genomic level, and quantifying genetic load, etc. (Allendorf et al. 2010, 2022; DeWoody et al. 2021). Genetic data can also detect gene flow between domesticated species, their wild relatives, and/or genetically modified organisms or improved varieties (Rojas-Barrera et al. 2019). The use of recently defined Genetic Essential Biodiversity Variables—standardized genetic summary metrics that focus on core attributes of genetic composition—can facilitate comparison among genetic monitoring

studies (Hoban et al. 2022). Genetic data can sometimes also provide information on the type and extent of local adaptation and maladaptation (Hoban et al. 2016; Capblancq et al. 2020). Combining genetic data with simulations can predict likely trajectories of populations and their fitness and diversity in response to management (Hoban 2014; Robinson et al. 2022) including time until a certain threshold of genetic erosion occurs (Hoban et al. 2014; Willoughby et al. 2015). This kind of information is often vital to on-the-ground management decisions as well as informing policy. As a few of the many examples, genetic data contributed to decisions about successful genetic rescue for helmeted honeyeaters in Australia (Harrisson et al. 2016; Pavlova et al. 2023), thresholds for hybrid management in bontebok and blesbok in South Africa (van Wyk et al. 2017), and red wolf management in the USA (Gese et al. 2015) (see also Bertola et al. 2024). Nonetheless, recent surveys show that a very small portion of species have any genetic data available (perhaps 1 to 5%) and an even smaller portion (less than 0.5%, even in wealthy nations) have temporal genetic monitoring (Torres-Florez et al. 2018; Posledovich et al. 2021; Thurffjell et al. 2022; Pearman et al. 2024; Paz-Vinas et al. 2023).

Synthesis

As discussed above, genetic indicators and genetic data collection methods both provide data necessary to monitor and protect genetic diversity, albeit in different ways. They are complementary strategies that should be deployed together for conserving and managing biodiversity. While genetic data can be used to generate detailed analyses of a given entity's genetic status, genetic data alone have not been sufficient to meet all conservation genetic needs, including prioritizing among many species, and supporting affordable and rapid decision-making. Conversely, while genetic indicators enable broad-scale evaluation of many species and facilitate affordable and accessible tracking, reporting, prioritization and communication, they are proxies and do not provide a full assessment of genetic composition and genetic change in a population or species, nor answer applied questions such as whether gene flow is occurring between locations. Genetic indicators (e.g., the proportion of large populations or the proportion of extant populations relative to extirpated populations; Hoban et al. 2023a, b) can be compiled for one species in just a few hours if field-based ecological or demographic data have already been generated, including via citizen scientists or local communities (Mastretta-Yanes et al. 2024a, b). In contrast, conducting an empirical genetic or genomic study commonly takes months, and substantial infrastructure, expertise, samples, and funding (Kriesner et al. 2020; Mastretta-Yanes et al. 2024a).

The complementary use of indicators and detailed genetic studies may be analogous to species threat assessments (e.g. IUCN Red List) and detailed population viability models (PVMs). Like genetic studies, PVMs offer a high level of precision and insight, although they typically require detailed information that is slow and expensive to gather, making their application feasible for relatively few species. In contrast, species threat assessments, like indicators, use pragmatic thresholds of a few simple criteria to characterize a species' status, enabling them to be done for many species, and indicating where more detailed studies are required. Like broad-scale threat assessments, genetic diversity indicators can point to potential issues, raising alarm in situations where concern is warranted, which can promote collection of more data, generate specific actions, and encourage monitoring.

Genetic indicators can be integrated with genetic data to provide a comprehensive genetic management approach (Fig. 1). For example, indicators may reveal which species or populations are most likely in need of genetic management, and genetic data can then be generated for those species to provide management-relevant information such as migration rates, temporal changes in N_e , and inbreeding. Meanwhile, DNA-based genetic studies of wild populations can provide better understanding of the biological processes underpinning genetic change, such as N_e dynamics, determinants of the N_e/N_c ratio, undesired effects of gene flow or artificial selection, and the needs of populations for long-term persistence. Such genetic knowledge can then be directly used to improve indicator calculation, such as by clarifying population genetic structure of a species or applying a tailored N_e/N_c ratio, leading to increasingly robust, empirically supported indicator estimates.

Conclusion

In summary, the development of simple indicator approaches enables tracking and reporting of genetic information at large scales in all countries. While genetic studies provide irreplaceable information often useful for on-the-ground management, especially for issues like hybridization, inbreeding, fitness, and adaptation, they are challenging to scale up and therefore should be complemented by less expensive and more scalable approaches (e.g., indicators). Both are important and useful for genetic conservation action and should be used to complement each other. We conclude that a holistic approach at a country-level through the development of both genetic indicators and genetic studies can rapidly improve the monitoring and conservation of genetic diversity.

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Author contributions SH drafted the initial manuscript, CG helped revise key points, RS and I P–V drafted the Figure, and all authors contributed edits, comments, or dialogue on the manuscript topics. All authors approved the final manuscript.

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Data availability No datasets were generated or analysed during the current study.

Declarations

Competing interests Sean Hoban is an editor for Conservation Genetics; there are no other conflicts of interest to declare.

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