Meetings

Genetics and genomics in wildlife studies: Implications for ecology, evolution, and conservation biology

Introduction

The meeting ''Genetics and Genomics in Wildlife Studies: Implications for Ecology, Evolution, and Conservation Biology'' was held in Seville, Spain, on October 20–21, 2011. The event highlighted the potential applications of cutting-edge genomic approaches for ecological, evolutionary, and conservation genetics research. The current revolution in next generation sequencing (NGS) technologies is destined to add a genomic dimension to biological diversity studies. Twelve leading researchers were invited to give talks on the application of genomic tools to address evolutionary, ecological, and conservation questions. Talks covered work currently being done using genomic approaches, either in species with a reference genome or in non-model organisms, to tackle a wide array of evolutionary questions. The event raised interesting novel insights into the applicability of genomics to conserve biodiversity.

Comparative genomics

The increasing number of species with available genomes has enabled comparative studies of genome structure, function, and evolution. Indeed, Warren Johnson (Center for Cancer Research, Bethesda, USA) announced that 200 new genome assemblies are about to be released, as part of the Genome 10K Project to sequence 10,000 vertebrate genomes. As an example of comparative approaches, Matthew Webster (Uppsala University, Sweden) presented his work on the *Prdm9* gene, known to play a key role in mammalian recombination. He found that this gene is not functional in the canid lineage. Scott Edwards (Harvard University, USA) has combined very different sources of information as part of his comparative genomics investigations of mammals, reptiles, and birds. His phylogenomic analyses of reptiles' bacterial artificial chromosomes (BACs) demonstrated a reduction of genome size from the common ancestor of amniotes through reptiles, culminating in tiny bird genomes. Additional analyses of osteocytes in dinosaur fossils, a proxy for genome size, showed an association between small genomes and flight.

The NGS revolution allows new integrative approaches combining comparative and population genomics. Rasmus Nielsen (University of Copenhagen, Denmark) emphasized how a single diploid genome can inform about ancient demographic events and assign population ancestry using appropriate analyses. Comparing the genome of one ancient human with single nucleotide polymorphisms (SNPs) in modern humans, he inferred that the *Saqqaq*, an extinct culture from Greenland, was related to North Eastern Siberian populations. Similarly, Mattias Jakobsson (Uppsala University) spoke about population genomics and human evolutionary history. He used a combination of simulations and principal component analyses (PCA) projecting extant human variation onto PC axes defined by Denisovan (a hominid from Siberia), Neanderthal, and chimpanzee genomes. He found that contemporary East Asian populations exhibited close genetic affinity with the Denisovan genome. Jakobsson also noted that the analysis of copy number variation (CNV) in the human genome is as important as SNP variation analyses. Tomàs Marquès-Bonet (Evolutionary Biology Institute UPF-CSIC, Barcelona, Spain) focused on ape comparative genomics and mapped CNVs and segmental duplications. Marquès-Bonet has sequenced 91 primate genomes and is using the human genome as a reference for reliable identification of CNVs. He also explained the efficiency of their protocols for detecting homozygous blocks in the genome of captive animals. For example, the study of ''Snowflake'', an albino captive gorilla, revealed a similar mutation to the one responsible for albinism in humans.

Adaptation genomics

The NGS explosion has promoted the use of genomics to study adaptation and selection in far more detail than previously possible, which has potential implications for conservation. Indeed, genome-wide selection scans often analyze the allele frequency spectrum to identify positive selection. Nielsen clearly explained how selective sweeps produce an excess of high frequency alleles in positively selected genes. For example, comparing whole exomes of 50 Tibetans and 40 Han Chinese samples, he detected a strong signal of selection on EPAS1, a gene possibly associated with high altitude adaptation in Tibetans. Additionally, regions

DOI 10.1002/bies.201100171

affected by a selective sweep typically show high levels of linkage disequilibrium (LD). Comparison of 40 light-coverage genomes of silkworm allowed Nielsen to identify a region of high LD surrounding the silk gland factor-1 (SGF-1).

Other genome scans frequently look for regions showing strong genetic differentiation. For example, Webster genotyped more than 170,000 SNPs in 46 dog breeds. High F_{ST} values between certain breeds indicated selective sweeps in particular genomic regions that could be associated with distinctive phenotypic traits. Similarly, William Cresko (University of Oregon, USA) used genotyping of high throughput restriction-site-associated DNA (RAD) tags to investigate genomic patterns of adaptation associated with transition from oceanic to freshwater environments in sticklebacks. His data showed how selection repeatedly targeted similar genomic regions associated with phenotypic adaptations to these aquatic environments showing how genomes might be shaped by ecological factors.

As an alternative to genome-wide scans, a focus on a more restricted set of candidate genes is still useful and also facilitated by NGS technologies. Robert Wayne (University of California, Los Angeles, USA) identified a gene responsible for black color in dogs that introgressed into North American wolves only a few thousand years ago. Follow-up studies showed how the black coat color polymorphism is maintained in wolves through trade-offs between lower fecundity and higher survival, possibly due to enhanced immunity. Victoria Sork (University of California) used a landscape genomics approach combining ecological, genetic, and spatial information to better understand how gene flow and selection shape diversity in Californian savanna oaks. She genotyped 194 SNPs in 50 genes likely related to environmental response. She found signals of climate adaptation by means of multivariate analyses corrected for spatial genetic structure. Following this trend, Craig Primmer (University of Turku, Finland) developed and analyzed 16 microsatellites linked to immune related genes in Atlantic salmon populations and found a negative relationship between genetic diversity and latitude. This is consistent with increasing pathogen-driven selection under higher temperatures. In the context of a global-warming scenario, the response of natural variation to climatic conditions is of conservation concern.

Although we can now identify some genes involved in the adaptation to a specific environment, natural selection acts on phenotypes. A first step toward linking genotype and phenotype could involve a proteomic or transcriptomic approach that does not require a reference genome. For example, Robert Ekblom (Uppsala University) used transcriptomic characterization to compare male mating behavior with associated plumage polymorphisms in the ruff, a lekking bird. Additionally, he genotyped SNPs identified from transcriptome sequences to search for genetic divergence due to mating strategies. Gene expression studies in different domestic chicken breeds (egg-layers and gamecocks) allowed Judith Mank (Oxford University, UK) to better understand the evolution of genes controlling sexual dimorphism. Her results showed that genes influencing sexual dimorphism are located on both sex chromosomes and autosomes. She also found how sex-biased gene expression and selection changed during development associated with periods of peak sex-hormone production.

Implications

The meeting ended discussing the status and upcoming needs of conservation genomics. A range of important needs were identified including: cost-effective genotyping methods at a population level, sequencing methods for low quantity or poor quality DNA, and the assembly of duplicated genomes (e.g. amphibian, fish, and plant species).

Data storage is becoming a demanding issue. The burst of whole genome data requires improvements in our storage strategies, such as format standardization and data exchange capacities through public virtual museums or commercial cloud computing platforms. Collaborative projects, such as the Genome 10K, are required to maximally represent phylogenetic diversity with the added benefit of genome-enabling closely related species. It is also necessary to enhance sequencing efforts including other large groups such as arthropods, fungi, and plants. This information would be invaluable for studying the evolution and conservation of extant biodiversity.

There is a continuing need for theoretical, statistical, and bioinformatic developments because of the huge amount of NGS information becoming available. For example, genome-wide association studies (GWAS) have had mixed success in accounting for the total heritability of traits or identifying genes of relatively small phenotypic effect. Future studies require new ways to combine information about genealogies, interactomics, gene regulation, and clear-cut phenotype characterization.

NGS is dramatically increasing our analytical power and accuracy. However, discussion at the end of the meeting cautioned against overuse of NGS if the same questions could be answered with less genetic data. We should carefully consider our research priorities and justification for a genomic approach when resources dedicated to conservation are limited. Genomics will definitely increase our understanding of adaptation and fitness components in a conservation context, but this might sometimes be overshadowed by the priority of preserving processes and habitats in the immediate short term for highly threatened species.

Acknowledgments

We thank fellow meeting organizers: Giulia Crema, José A. Godoy, Jennifer A. Leonard, and Carles Vilà. We are grateful to all invited speakers for their thought-provoking contributions. We also thank Foro de la Biodiversidad and EBD-CSIC staff for assistance. This meeting was financially supported by the *EcoGenes* project (Ref.: 264125) of the EU FP7-REGPOT-2010-1 program.

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