



EVOLUTION

Avian supergenes

Genetic data reveal how two complex bird mating systems evolved

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As the extravagant displays of birds of paradise remind us, many birds go to great lengths to pass their genes on to the next generation. Recent papers explore the genetic basis of the reproductive strategies in two bird species: the ruff (*Philomachus pugnax*) (1, 2) and the white-throated sparrow (*Zonotrichia albicollis*) (3). In each species, striking variation in both plumage and behavior is controlled in concert via the inheritance of coadapted gene complexes (supergenes) in large chromosomal inversions. The similar ways in which these independently evolved supergenes influence morphology and behavior elucidate how complex phenotypes evolve and are maintained.

Supergenes are two or more linked genes that are inherited together. They can play an

important role in generating biological diversity in taxa ranging from plants to humans. The mimetic wing patterns of butterflies are a particularly well-documented example of how supergenes can underlie complex adaptations (4). However, knowledge of the genetic architecture of supergenes remains limited, and the molecular mechanisms by which they can generate complex phenotypes are unclear. The recent studies of the ruff (1, 2) and the white-throated sparrow (3) provide critical advances to our understanding of these aspects of supergenes.

Reduced recombination within supergenes is central to their evolution, allowing multiple genes to be inherited as a single linked unit and setting the stage for their coevolution. Inversions, chromosomal rearrangements where large portions of the DNA molecule are flipped, suppress local recombination (5). Perhaps not surprisingly, inversions harbor supergene complexes (4) in groups that now include the ruff and the white-throated sparrow (1–3). In each of these bird species, individuals can exhibit strikingly different plumage and behavior that we can now link to genomic variation.

The ruff is a lek-breeding wading bird in wetlands of northern Eurasia (6). Male ruffs occur as one of three morphs, each with a distinct breeding strategy and appearance (see the photo). Independent males (80 to 95% of the population) are dominant holders of display sites, with diverse patterns of ornamented dark plumage. Satellite males (5 to 20%) are also ornamented, but with white plumage. They do not defend display sites, but rather visit the leks of independent males, where they assist with female

Lekking ruffs. From left to right, a female, an independent male, and a satellite male ruff. A third type of male ruff closely resembles the female. Recent research has shed light on the genetic underpinnings of the ruff's complex reproductive strategies (1, 2).

attraction and attempt to “steal” copulations. Faeder males (<1%) mimic females in their plumage and smaller size and also steal copulations.

As Küpper *et al.* (1) and Lamichhaney *et al.* (2) now show, these three reproductive behaviors and associated phenotypes are determined by a ~4.5 Mb inversion located on an autosome (see the figure). Independent males carry two copies of the ancestral, non-inverted chromosome: They do not possess either ruff supergene. Satellites and faeders have different supergenes: They each carry one ancestral haplotype and one morph-specific version of the supergene. Females carrying either version of the supergene can also be identified by size or from their behavioral response to testosterone implantation (1).

The white-throated sparrow is a common North American bird with two equally abundant color morphs: white-striped and tan-striped. White males invest in securing mates at the expense of parental care, whereas tan males are monogamous and help to look after their young (7). A similar trade-off is also seen in females. The two morphs are controlled by the presence of an inversion that harbors a large, ~100 Mb supergene, roughly 10% of the genome (3, 8, 9), and Tuttle *et al.* (3) have disentangled its evolutionary origin. White birds carry one copy of the supergene and one noninverted form, whereas tan birds only carry the non-

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inverted form (see the figure). White birds of either sex nearly always mate with tan birds of the opposite sex.

Although the supergenes of white-throated sparrows and ruffs are both ancient, they have different origins. The ruff supergenes arose from rearrangements in the ruff genome. The faeder supergene appears to have arisen ~3.8 million years ago via an inversion of the ancestral chromosome. The satellite supergene may be a product of rare recombination between the ancestral ruff chromosome and the faeder supergene that took place ~0.5 million years ago (2). In contrast, in white-throated sparrows, Tuttle *et al.*'s phylogenomic analyses indicate that the supergene predates the existence of that species and may be the product of introgression via hybridization with a now extinct relative (3).

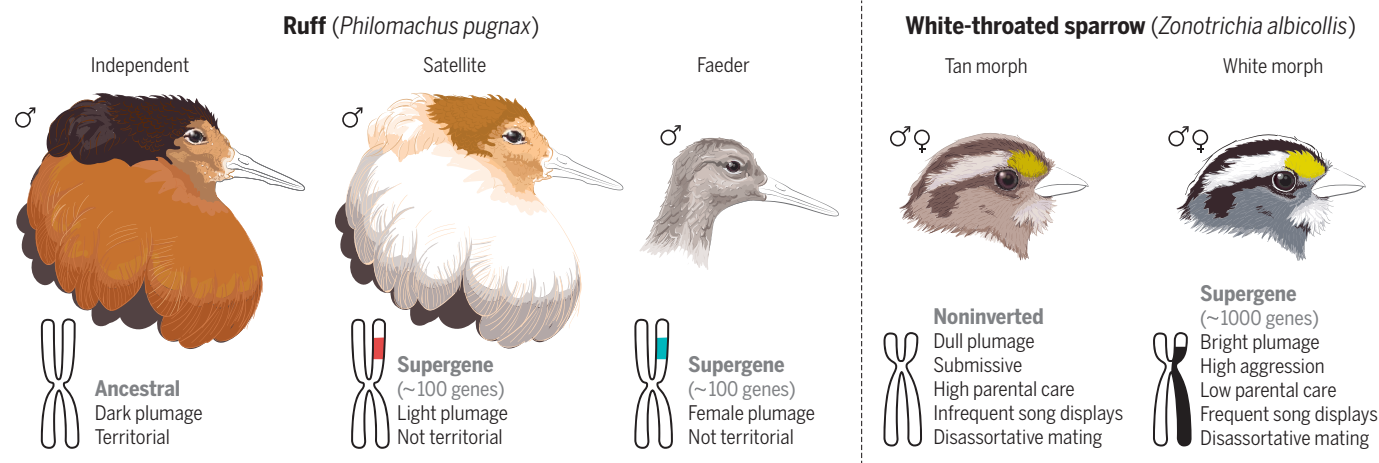
In other taxa, supergenes and their potentially adaptive gene complexes have spread via hybridization (4), but the relative importance of hybridization versus intraspe-

the precise mechanism remains unknown (3). Why do the supergenes persist despite these fitness costs? The answer is that each mating system allows alternative reproductive strategies to coexist as balanced polymorphisms. Satellite and faeder ruffs are hypothesized to have fitness advantages that maintain these morphs at low frequencies (1, 2). Polymorphism in sparrows persists via their mating pattern (white nearly always mates with tan, producing both tan and white offspring) and similar fitness benefits to either mating strategy (3).

Although the ruff and sparrow supergene variants can each be unambiguously linked to morph-specific traits, moving past this general association will be challenging. Each supergene region contains from 100 (ruff) to over 1000 (white-throated sparrow) genes, all of which are highly differentiated from their respective noninverted haplotypes (1–3). The divergent alleles on the supergenes could drive differentiation between morphs, yet there is evidence that the supergenes are

cases, loci involved in the response to sex hormones and plumage pigmentation exist in the supergene and likely act in concert to produce the alternative mating strategies and their associated behaviors and morphologies. In two closely related crow species, another putative inversion-generated supergene also contains genes involved in pigmentation and hormonal regulation (11). This mechanism is different from that seen in the rhinoceros beetle (*Trypoxylus dichotomus*), where differential sensitivity to growth hormones based on a single signaling gene has been proposed as a molecular mechanism by which ornaments and weapons can grow to exaggerated proportions (12).

Considered in concert, the findings in ruff and white-throated sparrow highlight the importance of supergenes in the generation of complex phenotypes. They provide insight into the varied ways in which supergenes can be generated and maintained and begin to expose their genetic architecture. These recent studies provide a link between genotype and



The genetic architecture of two avian mating systems. The presence of inversion-generated supergenes determines morphological and behavioral traits in ruffs and white-throated sparrows. The ruff supergenes harbor close to 100 genes; the white-throated sparrow supergene is much larger, with over 1000 genes.

cific chromosomal rearrangements for the generation of supergenes is not known. The recent studies (1–3) show that supergenes with similarly complex influences on morphology and behavior can arise in both ways. It remains to be shown whether supergenes tend to be the product of within-species processes, or are more often co-opted from other species via introgressive hybridization.

In both birds, there are fitness consequences associated with possessing the supergene, yet the polymorphisms appear demographically stable. The ruff supergenes are lethal when either sex has a copy on each chromosome (1), whereas the sparrow supergene only appears to be lethal when males have a copy on each chromosome (3). In ruffs, the inversion interrupts a gene essential for mitosis (1, 2); in sparrows,

degrading (1–3), as is the case for the mammalian Y and avian W sex chromosomes (10).

Typically, experimental crosses can be used to tease apart regions of the genome involved in generating a trait. However, the lack of recombination within supergenes makes this an unfeasible approach. Now that the genes within the ruff and white-throated sparrow supergenes are known and the genomic resources for these nonmodel organisms have been generated, the relative influence of different genes in each supergene can be explored with targeted gene expression analyses and genome-editing technology.

The broad overlap in the function of the genes within the ruff and sparrow supergenes provides a hint as to how their complex phenotypes could be generated. In both

phenotype, a long-standing goal in evolutionary biology research. Supergenes may have an underappreciated role in the generation of biodiversity, one that we are now poised to more fully appreciate and understand. ■

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