Supergenes are clusters of physically linked, co-evolving genes that often control complex traits. A new study clarifies the origin and possible fate of a fascinating supergene that determines the coloration and mating behavior of a widespread North American bird.

Genetic variation in natural populations leads to differences among individuals. Some variable traits have a relatively simple genetic basis; think, for example, of Mendel’s famous peas. In contrast, the generation of a complex trait like human height is generally less straightforward, as multiple interacting genes control it. Understanding the genomic architectures that generate complex phenotypes is a central goal in current evolutionary biology research. Elaborate traits are often produced through the coordinated effect of many genes, and require fine-tuned regulatory networks for gene products to interact in the appropriate tissue at the right time. The evolution of traits generated by numerous genes can be modeled by assuming that they are controlled by large numbers of genes scattered around the genome, each contributing additively to the final phenotype [1].

How do independent genes evolve to act together and produce traits like elaborated ornaments or complex behavioral patterns? A possibility is that instead of being scattered across the entire genome, interacting genes are in fact clustered together within a region of a chromosome (i.e., physically linked). Such so-called ‘supergenes’ could then be inherited as a single unit and potentially regulated in concert [2]. Chromosomal inversions — structural rearrangements in which large portions of a chromosome are flipped — are one mechanism by which supergenes may arise and evolve [3]. Inversions disrupt the pairing of chromosomes during meiosis, and single crossing over events within the inversion result in inviable gametes with extra or missing genes [3]. As a result, recombination within the inversion is vastly reduced in individuals heterozygous for the rearrangement, and the genes within it become tightly linked. Complex phenotypes determined by supergenes include mimicry in butterflies, colony organization in eusocial insects, and the fecundity of women in a particularly well-studied Icelandic population (reviewed in [2]). In many organisms with chromosomal sex determination, the differences between the sexes themselves are traits controlled by a supergene: the non-recombining portions of the sex chromosomes [2]. In a new paper in this issue of Current Biology, Tuttle et al. [4] use an impressive wealth of genomic and long-term field data to study a massive supergene that controls both morphology and behavior in the White-throated Sparrow (Zonotrichia albicollis).

The White-throated Sparrow is a widespread North American bird that is common in backyards and at birdfeeders. A close look reveals that it has two color morphs occurring in similar proportions: white-striped and tan-striped (Figure 1A,B). Remarkably, white birds of either sex mate almost exclusively with tan birds of the opposite sex [4,5], a pattern which results in four effective ‘sexes’, as any given individual can breed with only one-quarter of the population (Figure 1C). White and tan individuals also show alternative reproductive strategies [6]: white males are aggressive, provide little parental care, and are promiscuous, whereas tan males are monogamous and invest highly in rearing their nestlings. White females are more aggressive and solicit more copulations from their mates than do tan females [6]. Reproductive tradeoffs between colorful and dull morphs have been observed in other bird species [7], but in this sparrow the long-term reproductive success achieved through either strategy is similar [4]. The white and tan morphs are controlled by a series of nested inversions on chromosome 2 encompassing ~100 megabases [4,5] (Figure 1D). This extraordinarily large supergene constitutes ~10% of the entire White-throated Sparrow genome [8].

White birds are heterozygotes for the inverted chromosome 2/2m; ‘m’ stands for metacentric and describes the position of the centromere), while tan birds do not carry the inversion (2/2). Because white sparrows almost never breed with similarly heterozygous white mates, offspring with two copies of 2m are rarely produced. The supergene captures ~1,100 known genes, many of which are candidates for morph-specific behavior [9] and plumage [4]. The inverted portions of 2 and 2m are extremely divergent (~1 fixed difference every 100 base-pairs), leading researchers to hypothesize that 2m may have had an ancient origin [5,8,10]. Tuttle et al. [4] explain the origin of 2m by using whole-genome sequences to build a phylogeny for the Zonotrichia genus and a few close relatives. In contrast to the genome-wide evolutionary relationships, the phylogenetic tree derived from the 2m supergene suggests an extremely ancient split for only this subset of the sparrow genome, originating long before the White-throated Sparrow split from its closest relative. The authors interpret this finding as evidence that 2m originated in an extinct species and then spread into the White-throated Sparrow through ancient hybridization (such an explanation has been suggested for other supergenes in different species [2]).
Are the divergent genes on 2\textsuperscript{m} responsible for generating the white morph or is this phenotype a consequence of genes on 2 being represented by a single chromosome in white (2/2\textsuperscript{m}) individuals? Theory predicts that selection should be inefficient at purging deleterious mutations on the 2\textsuperscript{m} supergene because it nearly always exists in a heterozygous, non-recombining state [11], and genes on 2 could be able to compensate if genes on 2\textsuperscript{m} become non-functional. This leads to the prediction that the 2\textsuperscript{m} supergene should degrade genetically, analogously to the mammalian Y and avian W sex chromosomes [11]. In contrast to this expectation, examination of some genes within the 2\textsuperscript{m} supergene does not support this hypothesis [9,10]. Individuals homozygous for 2\textsuperscript{m} are extremely rare (Tuttle et al. [4] found only 3 out of 1989 pairs documented by Tuttle et al. [4] in a 27-year study. (D) A model for the series of inversions that led to chromosome 2\textsuperscript{m} (reproduced from [10]). The circle represents the centromere and arrows indicate relative orientation. Dashed lines show the hypothesized breaking points that led to the inversion and ‘?’ denotes a possible intermediate state. Recombination between 2 and 2\textsuperscript{m} only occurs on the red portion of the chromosome. Illustrations by Liz Clayton Fuller.

2\textsuperscript{m} degradation is needed. Accumulation of transposable elements and mutations that render proteins non-functional will answer the question of whether or not 2\textsuperscript{m} is indeed degrading like a Y or W sex chromosome.

New genomic techniques are helping discover other fascinating examples of elaborate phenotypes and divergent reproductive strategies controlled by supergenes in birds [15,16]. The exact connection between the molecular mechanisms and the traits — including the nearly perfect disassortative mating in the White-throated Sparrow — remains unknown. Yet Tuttle et al. [4] have moved a substantial step further in this direction. What is now certain is that while every field guide in North America refers to this sparrow species as ‘common’, its genetics suggest otherwise.

REFERENCES

Plant Physiology: Redefining the Enigma of Metabolism in Stomatal Movement

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Stomata open at the leaf epidermis, driven by solute accumulation in the surrounding guard cells. Transmembrane ion transport has long been recognised to contribute to this process. A new study makes it clear that guard cells also metabolise starch to accelerate opening.

Stomata are pores that provide the major route for gaseous exchange across the impermeable cuticle of leaves and stems. Stomata open and close to facilitate CO₂ entry to the interior of the leaf while controlling water loss by transpiration. They exert major controls on the water and carbon cycles of the world [1] and are arguably at the centre of the crisis in water availability and crop production that is expected to unfold over the coming decades. Guard cells occur in pairs surrounding the stomatal pore and respond in a well-defined manner to an array of extracellular signals — including light, relative humidity and CO₂ — to regulate stomatal signals, including light, relative humidity and CO₂, to regulate stomatal opening.

Ironically, while much research between the 1960s and 1980s was motivated by a desire to understand what makes stomata open, it was soon eclipsed by interest in what makes them close. In many respects, the process of solute accumulation in guard cells for stomatal opening is similar, if not identical, to that for cell expansion and growth. Stomatal closing was seen to be different, however — very few plant cells reversibly increase and decrease in cell volume. MacRobbie’s pioneering radiotracer flux studies on the effects of the water-stress hormone abscisic acid [2] and evidence of its coordinated actions on guard cell K⁺ channels [3,4] left no doubt that stomatal closure was a concerted and highly regulated process. Research soon established the mechanisms of transport and its regulation during stomatal closing, accelerated by the combined technologies of the voltage clamp and molecular genetics [5], and subsequently by those of Arabidopsis genomics [6,7] and systems biology [8,9]. However, knowledge of stomatal opening and its integration with metabolism languished far behind.

A handful of studies indicated that sugar and malate (Mal) accumulated in guard cells of open stomata and suggested that guard cells could mobilise starch to generate organic osmotica. Talbott and Zeiger [10] observed substantial increases in sucrose (and under blue light also Mal) that correlated with stomatal aperture in Vicia. MacRobbie, too, found that inorganic solute could not account fully for the osmotic content of guard cells of open stomata in intact leaves of Commelina [11]. However, conflicting data from other species failed to yield any consensus. Even in Vicia, the accumulation of Mal was known to depend on the availability of Cl⁻ as a counter-ion for K⁺ [12,13]. Thus, in the minds of many, myself included, the production of organic solutes was secondary, a sideshow that contributed osmotica if sufficient inorganic solute was not available.

In a report published in this issue of Current Biology, Horrer et al. [14] effectively turn this viewpoint on its head with a study that will energize research...