

COMPLEX TRAITS

A diamond in the ruff

“ a simple genetic factor, a so-called supergene, can underlie the evolution of multiple complex traits ”

Two new studies of phenotypic variation in male ruffs (*Philomachus pugnax*), a sandpiper species with three genetically determined types of males, independently show that a simple genetic factor, a so-called supergene, can underlie the evolution of multiple complex traits.

Ruffs are wading birds with three male morphs, that is, cohabiting animals of the same species that differ markedly in their appearance and mating behaviour: the territorial ‘independents’ have dark ornamental plumage, which they use to attract and court females; the less frequent ‘satellites’ have white plumage and are non-territorial but team up with a territorial male and attempt to mate when it is distracted; and the rare ‘faeders’ physically resemble females to avoid detection by other males and attempt to slip between females and ornamented males to mate quickly.

Lamichhaney *et al.* sequenced the genomes of wild ruffs and identified a single 4.5 Mb region that was highly differentiated between independents and satellites. In phylogenetic trees, independents and satellites clustered separately based on this region only (compared with trees based on the rest of the genome). Structural variant analysis revealed an inversion between independents and satellites that localized precisely to the highly differentiated region. Sequence comparison between ruffs and chickens

indicated that the allele in independents was the ancestral, non-inverted version. Analysis of the patterns of differentiation within this region among all three morphs suggested that the inversion arose around 3.8 million years ago, giving rise to an ancestral *Faeder*-like allele, and that the *Satellite* allele arose around 500,000 years ago from a rare recombination event between the non-inverted *Independent* and inverted *Faeder*-like alleles.

Küpper *et al.* used a different approach that included ruffs from their own pedigree captive breeding population as well as wild ruffs. The authors carried out restriction site-associated DNA (RAD) sequencing to identify single-nucleotide polymorphisms (SNPs) in their pedigree population, and used these to map the *Faeder* and *Satellite* alleles to the same genomic region. Linkage analysis suggested that this region was refractory to recombination between haplotypes. Based on SNP data from unrelated wild ruffs, the authors determined that specific segments within this region were uniquely associated with each morph. To fine map this variation, they sequenced the genomes of five wild males and assessed nucleotide variation across the region between morphs. The inversion was identified based on nucleotide divergence and on the orientation of read pairs at the inversion breakpoints. Some regions of the satellite inversion

were more similar to independents than to faeders, consistent with the *Satellite* allele having arisen from a rare recombination event between the inverted *Faeder*-like and non-inverted *Independent* alleles, as proposed by Lamichhaney *et al.*

Both groups report the disruption of the essential *CENPN* gene (encoding centromere protein N) at the breakpoint of the inversion, which should result in recessive lethality. Accordingly, pedigree data from the study by Küpper *et al.* confirmed a total lack of homozygotes for the inversion. The two studies identified multiple missense mutations and deletions within the inverted region in satellites and faeders, many of which were in or near genes associated with hormone systems, potentially explaining the differences in reproductive traits between morphs. Interestingly, both groups identified mutations in the *MC1R* gene, which is associated with plumage colour, as a possible explanation for the white colour of satellites.

The authors of these studies propose that, by suppressing recombination, the inversion created a ‘supergene’ in which adaptive changes accumulated over millions of years, giving rise to the complex phenotypic and behavioural differences between morphs.

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ORIGINAL ARTICLES Lamichhaney, S. *et al.* Structural genomic changes underlie alternative reproductive strategies in the ruff (*Philomachus pugnax*). *Nat. Genet.* <http://dx.doi.org/10.1038/ng.3430> (2015) | Küpper, C. *et al.* A supergene determines highly divergent male reproductive morphs in the ruff. *Nat. Genet.* <http://dx.doi.org/10.1038/ng.3443> (2015)



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