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京都大学農学部総合館 W-100講義室

Genetic characterization of a supergene controlling male mating strategies in ruff

A paradox in evolutionary biology is how supergenes can maintain high fitness despite reduced effective population size, the suppression of recombination, and the expected accumulation of mutational load. The ruff supergene involves two rare inversion haplotypes (*Satellite* and *Faeder*) (Lamichhaney et al. 2016). These are recessive lethals but with dominant effects on male mating strategies, plumage, and body size. Sequence divergence to the wild-type (*Independent*) haplotype indicates that the inversion could be as old as 4 million years. We have constructed a highly contiguous genome assembly of the inversion region for both the *Independent* and *Satellite* haplotypes. Based on the new data we estimate that the recombination event(s) between an inverted and non-inverted chromosome creating the *Satellite* haplotype occurred only about 70,000 years ago. Contrary to expectations for supergenes, we find no substantial expansion of repeats and only a very modest mutation load on the *Satellite* and *Faeder* haplotypes despite high sequence divergence to the non-inverted haplotype (1.46%). The essential centromere protein N gene *CENPN* is disrupted by the inversion, and surprisingly is as well conserved on the inversion haplotypes as on the noninversion haplotype. These results suggest that the inversion may be much younger than previously thought. The low mutation load, despite recessive lethality, may be explained by the introgression of the inversion from a now extinct lineage. We also observe accelerated evolution of the *melanocortin-1 receptor* gene (*MC1R*) which shows four missense mutations between the *Satellite* and *Independent* alleles which is likely to cause light colored ornamental feathers in *Satellite* males. *MC1R* mutations are causing pigmentation variation in chicken, cattle, horse, pigs and other domestic animals.

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