New study reveals how some chickens got striped feathers

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Coucou de Rennes, a French breed with the characteristic sex-linked barring phenotype. Credit: Hervé Ronné, Ecomusée du pays de Rennes

Birds show an amazing diversity in plumage colour and patterning. But what are the genetic mechanisms creating such patterns? In a new study published today in *PLOS Genetics*, Swedish and French researchers report that two independent mutations are required to explain the development of the sex-linked barring pattern in chicken. Both mutations affect the function of CDKN2A, a tumour suppressor gene associated with melanoma in humans.

Research in pigmentation biology has made major advances the last 20 years in identifying genes controlling variation in pigmentation in mammals and birds. However, the most challenging question is still how colour patterns are genetically controlled. Birds are outstanding as regards the diversity and complexity in colour patterning. The study published today has revealed the genetic basis for the striped feather characteristic of sex-linked barring. One example of this fascinating plumage colour is the French breed Coucou de Rennes. The name refers to the fact that this plumage colour resembles the barring patterns present in the common cuckoo (Cuculus canorus). The sex-linked barring locus is on the Z chromosome. (In chickens as well as in other birds the male has chromosomes ZZ while females have ZW).

"Our data show that sex-linked barring is caused by two independent mutations that act together. One is a regulatory mutation that increases the expression of CDKN2A. The other changes the protein sequence and makes the protein less functionally active. We are sure that both mutations contribute to the sex-linked barring pattern because we have also studied chicken that only carry the regulatory mutation and they show a very pale plumage with only weak dark stripes. Thus, this represents an evolutionary process in which the regulatory mutation occurred first followed by the mutation affecting the protein structure. The combined effect of the two mutations causes an even more appealing phenotype for the human eye," says Leif Andersson, Uppsala University, Swedish University of Agricultural Sciences and Texas A&M University, who led the study.

"The most important reason for the extensive colour variation among our domestic animals is that we appreciate this diversity, as long as the mutations underlying the variation are not causing health issues for the animals," says Leif Andersson.

The study illustrates how useful domestic animals are as models for evolutionary processes in nature. Leif Andersson argues that a similar evolution of gene variants comprising multiple genetic changes affecting the function of a single gene is the rule rather than the exception in natural populations.

CDKN2A is a well-studied tumour suppressor gene that takes part in the regulation of cell division and

cell survival. Mutations that inactivate CDKN2A are the most common explanation for familiar forms of melanomas in humans. (However, the great majority of melanoma cases are not associated with a strong genetic risk factor.)

"The gene variant underlying sex-linked barring has an opposite effect compared with the mutations causing melanoma in humans. Sex-linked barring is associated with a gene variant that makes CDKN2A more active leading to a cyclic deficit of pigment cells causing the white stripes during the development of an individual feather. It appears that pigment cells are particularly susceptible to changes in the function of CDKN2A as inactivating mutations in humans are associated with melanoma but rarely other cancer forms and activating mutations cause sex-linked barring in chickens but no other side effects are known," says Doreen Schwochow Thalmann, PhD student and first author of the paper.

"It is fascinating that a large proportion of chickens used for egg and meat production around the world carry these mutations in a tumour suppressor gene. An example of such a breed is White Leghorn which is one of the most prominent breeds used for egg production, but sex-linked barring is not apparent in these breeds because they also carry the dominant white colour that eliminates all pigment production and masks the effect of sex-linked barring," says Leif Andersson.

More information: Doreen Schwochow Thalmann et al, The evolution of Sex-linked barring alleles in chickens involves both regulatory and coding changes in CDKN2A, *PLOS Genetics* (2017). DOI: 10.1371/journal.pgen.1006665

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