Germinal reversion of the merle allele in Australian shepherd dogs

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ABSTRACT: A homozygous merle Australian shepherd bitch produced two nonmerle offspring and 64 that carried the merle allele. The two nonmerle offspring produced no merles when bred to nonmerles, indicating that the germinal reversion to nonmerle was stable. This occurrence is further evidence that the merle allele is due to a transposable DNA element.

The review of Whitney and Lamoreux concerning transposable elements of mammals indicates that perhaps the merle phenotype of dogs is due to one of these elements. Partial proof of this would be a high rate of reversion of germ cells from the mutant (merle) type to the normal (full colored) type. These reversions should be stable. Schable and Schable and Brombaugh have reported such reversions, which they termed mutations, from merle in a male collie, male Great Dane, and a female sheltie. The present report documents stable germinal reversions from merle in Australian shepherd dogs.

A homozygous merle Australian shepherd bitch produced 66 pups by nonmerle sires. The pups included 64 heterozygous merles and two (one male and one female) nonmerles. That the bitch is homozygous and not heterozygous is indicated by the deviation of the ratio from the 1:1 ratio expected of heterozygous merle by nonmerle crosses ($P = 2.970 \times 10^{-17}$).

The nonmerle bitch pup when bred to nonmerle males produced seven nonmerle pups. The nonmerle male pup produced four nonmerle pups by a nonmerle bitch, and when bred to heterozygous merle bitches produced no homozygous merles, eight heterozygous merles, and seven nonmerles. While a remote possibility, it must be considered that an occasional merle puppy with extensive full colored areas could be misclassified as nonmerle. However, the reversions cited here are not heterozygous merles, because their progeny differ significantly from heterozygous expectation (nonmerle bitch X nonmerle, $P = 0.008$; nonmerle male X nonmerle, $P = 0.0625$ (not significant); nonmerle male X heterozygous merle, $P = 0.0134$). They produce pups as though they were stable reversions to nonmerle.

Schable's reversions were produced by a male collie and a male Great Dane. The reversions in the Australian shepherd dog originated in a female. The rate of reversion in the collie was 1/21; in the Great Dane, 1/58; in the Australian shepherd, 2/66. The reversions are too few to consider that an occasional merle puppy with nonmerle type. These reversions should be stable. Schable and Schable and Brombaugh have reported such reversions, which they termed mutations, from merle in a male collie, male Great Dane, and a female sheltie. The present report documents stable germinal reversions from merle in Australian shepherd dogs.

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The difference between the merle (collie and Australian shepherd) and the harlequin merle (Great Dane) patterns are not known. The Great Dane produced merles in addition to harlequin merles. Schaible presents evidence that the two are due to different alleles at the same locus. This evidence comes from the breeding records of a composite heterozygous merle/harlequin merle sheltie. If this interpretation is correct then the nonmerle puppy described from this sheltie could be attributed to a crossover at the correct place in the allele, and may not be due to a germinal reversion from excision of a transposable element. Further characterization of the differences between merle and harlequin merle should help to choose between these alternatives.

NOTE ADDED: Since submission of this manuscript a homozygous merle daughter of the bitch described has produced 1 black and 9 merle pups. This brings the rate of reversion in this family of dogs to 3/76. The reversion in this individual has not yet been tested for stability, but it phenotypically resembles the ones that have been so tested.

References