Allelic Heterogeneity at the Equine KIT Locus in Dominant White (W) Horses

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White coat color has been a highly valued trait in horses for at least 2,000 years. Dominant white (W) is one of several known depigmentation phenotypes in horses. It shows considerable phenotypic variation, ranging from similar to 50% depigmented areas up to a completely white coat. In the horse, the four depigmentation phenotypes roan, sabino, tobiano, and dominant white were independently mapped to a chromosomal region on ECA 3 harboring the KIT gene. KIT plays an important role in melanoblast survival during embryonic development. We determined the sequence and genomic organization of the similar to 82 kb equine KIT gene. A mutation analysis of all 21 KIT exons in white FranchesMontagnes Horses revealed a nonsense mutation in exon 15 (c. 2151C > G, p. Y717X). We analyzed the KIT exons in horses characterized as dominant white from other populations and found three additional candidate causative mutations. Three almost completely white Arabians carried a different nonsense mutation in exon 4 (c. 706A > T, p. K236X). Six Camarillo White Horses had a missense mutation in exon 12 (c. 1805C > T, p. A602V), and five white Thoroughbreds had yet another missense mutation in exon 13 (c. 1960G > A, p. G654R). Our results indicate that the dominant white color in Franches- Montagnes Horses is caused by a nonsense mutation in the KIT gene and that multiple independent mutations within this gene appear to be responsible for dominant white in several other modern horse populations.
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