ABSTRACT

A mutation creating a potential illegitimate microRNA target site in the myostatin gene affects muscularity in sheep

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Texel sheep are renowned for their exceptional meatiness. To identify the genes underlying this economically important feature, we performed a whole-genome scan in a Romanov ×Texel F2 population. We mapped a quantitative trait locus with a major effect on muscle mass to chromosome 2 and subsequently fine-mapped it to a chromosome interval encompassing the myostatin (*GDF8*) gene. We herein demonstrate that the *GDF8* allele of Texel sheep is characterized by a G to A transition in the 3' UTR that creates a target site for *mir1* and *mir206*, microRNAs (miRNAs) that are highly expressed in skeletal muscle. This causes translational inhibition of the myostatin gene and hence contributes to the muscular hypertrophy of Texel sheep. Analysis of SNP databases for humans and mice demonstrates that mutations creating or destroying putative miRNA target sites are abundant and might be important effectors of phenotypic variation.

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