New kind of mutation is reported

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Belgium scientists say they have discovered a new kind of mutation that might be at the origin of many phenotypes in various species.

The researchers, led by University of Liege Professor Michael Georges, used a study on genetic factors that promote muscular hypertrophy among Texel sheep.

They report discovering a novel class of mutations that disrupt the function of a gene and cause a specific phenotype. The mutation created the appearance of an "illegitimate" microRNA recognition site in a gene that did not have it in its normal form.

In the study, the gene concerned is the myostatin, which is expressed in the skeletal muscle and the function of the derived protein is to inhibit muscular growth.

The mutation discovered among sheep exposed a recognition site for two microRNAs that are highly expressed in the muscle. In "mutant" animals, those microRNAs will consequently target the myostatin gene and block its translation.

The result, said the researchers, is that the absence of myostatin provokes a muscular hypertrophy among Texel sheep. But pursuing the study using bioinformatic approaches, the team identified polymorphisms among humans and mice that are likely to act in the same way.

The study appears in Nature Genetics.