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News Release

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Cell insights shed light on how muscle-wasting disease takes hold

Fresh insights into how our cells control muscle development could aid understanding of muscular dystrophy and other inherited diseases.

Scientists have discovered a way in which proteins in our cells help to control genes in our DNA, which are involved in forming muscle.

Their finding explains an apparent paradox in which proteins linked to a series of genetic diseases can be found in cells throughout the body, but impact only on some tissue types.

Researchers from the University of Edinburgh studied newly discovered muscle-specific proteins, found in a membrane that surrounds the DNA in each of our cells – known as the nuclear membrane. Previous studies have linked other proteins in this membrane to inherited diseases that affect only some tissues.

They used gene analysis technologies and microscopes to determine how these proteins are able to change the physical position of important genes during muscle development. They discovered that this re-locating can regulate the genes' ability to form muscle.

When researchers blocked the proteins from functioning, muscle could not be formed, demonstrating the extent of the proteins' direct control.

Researchers say the newly found level of gene control is likely to apply to all tissues. It is expected to have implications for other tissue-specific nuclear membrane conditions, such as fat, skin, bone and brain diseases, cardiomyopathy and an ageing disease known as progeria.

Their study, published in *Molecular Cell*, was funded by the Wellcome Trust.

Dr Eric Schirmer, of the University of Edinburgh's School of Biological Sciences, who led the study, said: "These results help to resolve some of the biological pathways behind nuclear membrane diseases. They also settle a paradox and resolve a debate among scientists, by showing how proteins found in all our cells can impact on some tissues, but not all – through interactions with tissue-specific proteins which, for most tissue types, await discovery."

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