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Cell discovery could hold key to causes of inherited diseases

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Fresh insights into the protective seal that surrounds the DNA of our cells could help develop treatments for inherited muscle, brain, bone and skin disorders.

Researchers have discovered that the proteins within this coating – known as the nuclear envelope – vary greatly between cells in different organs of the body.

This variation means that certain disease causing proteins will interact with the proteins in the protective seal to cause illness in some organs, but not others.

Until now scientists had thought that all proteins within the nuclear envelope were the same in every type of organ.

In particular the finding may provide insights into a rare muscle disease, Emery-Dreifuss muscular dystrophy.

This condition causes muscle wastage and heart problems, affects only muscles, even though it is caused by a defect in a nuclear envelope protein found in every cell in the body.

Scientists say that the envelope proteins they have identified as being specific to muscle may interact with the defective nuclear envelope protein that causes Emery-Dreifuss muscular dystrophy, to give rise to the disease.

In a similar way, this may help to explain other heritable diseases that only affect certain parts of the body despite the defective proteins being present in every cell. The study also identified nuclear envelope proteins specific to liver and blood.

Some of these also interact with proteins in all cells that are responsible for other nuclear envelope diseases, ranging from brain and fat to skin diseases, and so may help explain why things go wrong.

Dr Eric Schirmer, of the University of Edinburgh's Wellcome Trust Centre for Cell Biology, who led the study said: "Nobody could have imagined what we found.

The fact that most proteins in the nuclear envelope would be specific for certain tissue types is a very exciting development. This may finally enable us to understand this ever-growing spectrum of inherited diseases as well as new aspects of tissue-specific gene regulation."

The findings build on previous research that showed proteins in the nuclear envelope are linked to more than 20 heritable diseases.

More information: The study is published in the journal *Nucleus*.

Provided by University of Edinburgh