Generation of a human induced pluripotent stem cell line (CRICKi021-A) from a patient with Ullrich congenital muscular dystrophy carrying a pathogenic mutation in the COL6A1 gene

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New method to grow muscles in a dish to study UCMD

Researchers from the MAGIC project have developed a new laboratory tool to help study Ullrich congenital muscular dystrophy (UCMD), a rare genetic condition that affects muscles and connective tissue. This condition is caused by changes in certain genes, most commonly COL6A1, which is important for keeping muscles strong and stable.

Currently, there are no approved treatments for UCMD. One reason is that researchers still don't fully understand how the condition affects the body. Another challenge is finding reliable ways to test whether potential treatments might work.

To help solve this, the MAGIC project aims to create better models to study rare neuromuscular conditions, like UCMD, outside of the body, in the lab.

What is a lab model?

A lab model is a way for scientists to study a disease outside the human body - usually using cells grown in a dish. These models are designed to closely resemble the condition, helping researchers explore how it works and how it might be treated.

A new model for UCMD

In this study, scientists took small skin samples from people with UCMD who had a change in the COL6A1 gene. They used advanced technology to turn those skin cells into induced pluripotent stem cells (iPSCs). These special cells can grow into almost any type of cell in the body - including muscle cells.

This research gives scientists a new way to study UCMD in the lab and could help them:

- Understand how the condition affects muscle cells
- Track how it progresses over time
- Test new drugs or gene therapies
- Move closer to personalized treatments for people living with UCMD

Looking ahead

As part of the MAGIC project, researchers have now used this method to grow 3D mini-muscles – tiny, lab-grown muscle tissues that more closely look and behave like real muscles. These mini-muscles allow scientists to study UCMD in even more detail and test potential treatments in a way that better reflects what happens in people living with the condition. This can help identify which treatments show the most promise and should be prioritised for clinical trials.

Importantly, the researchers have shared the method for making these cells, so other scientists around the world can use it too. This kind of open collaboration helps speed up research and brings us all closer to finding effective treatments.

For more detailed information, you can refer to the full research article published in Stem Cell Research:
https://www.sciencedirect.com/science/article/pii/S1873506124003465