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Human Myotonic Dystrophy Induced Pluripotent Stem Cell Lines

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Life Sciences/Human Health
Life Sciences/Research Tools

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Myotonic dystrophy 1 (DM1) iPS cells

Primary fibroblasts from two DM 1 patients (DM1-1 and DM1-2) were reprogrammed in pluripotent cells by transducing with Oct3/4, Klf4, Sox2 and C-Myc. These DM1-1 and DM1-2 iPS cells were validated for pluripotent markers/capabilities and for the presence of DM1 RNA foci. Expansion of CTG trinucleotide repeats at the DMPK gene was also verified.

Organism: *Homo sapiens, human*
Tissue: Skin Fibroblast
Disease: Myotonic dystrophy patient
Karyotype: 46XY and 46XX [20/20 normal metaphase spreads]
Product format: Frozen
Applications: These cells can be grown in culture to model human disease. They can grow indefinitely and can be differentiated into myotubes. These cells are suitable for use as drug discovery and screening tools.

Myotonic dystrophy 1 (DM1) iPS cells expressing PAX7

Primary fibroblasts from two DM 1 patients (DM1-1 and DM1-2) were reprogrammed into pluripotent cells by transducing with Oct3/4, Klf4, Sox2 and C-Myc. The DM1-1 and DM1-2 iPS cells were modified with doxycycline inducible myogenic factor PAX7 (iPAX7). These DM1-1 and DM1-2 iPAX7 were differentiated into myotubes expressing DM1 RNA foci with evidence of sequestration of MBNL1.

Organism: *Homo sapiens, human*
Tissue: Skin Fibroblast
Disease: Myotonic dystrophy patient
Karyotype: 46XY and 46XX [20/20 normal metaphase spreads]
Product format: Frozen
Applications: These cells can be grown in culture to model human disease. They can grow indefinitely and can be differentiated into myotubes in vitro. These cells are suitable for use as drug discovery and screening tools.

Desired Partnerships

These cell lines are fully developed and available for license. Please contact our office to learn more.

Researchers

- **Rita Perlingeiro, PhD** Professor, Department of Medicine

References

1. Darabi, R., Arpke, R.W., Irion, S., Dimos, J.T., Grskovic, M., Kyba, M. and Perlingeiro, R.C.(2012) , https://doi.org/10.1016/j.stem.2012.02.015, Cell Stem Cell