Myotonic Dystrophy FOUNDATION

iPSC – Myotonic Dystrophy Animal Models & Tools

Animal models play a key role in basic, translational and clinical research. The following tables highlight and summarize available animal models and tools for myotonic dystrophy research. Literature links connect to the original publication.

This table summarizes available **induced pluripotent stem cells or cell lines (iPSC)** for myotonic dystrophy (DM). Such cell lines are derived from skin or blood cells that have been reprogrammed back into an embryonic-like pluripotent (undifferentiated or not yet committed) state that enable the development of an unlimited source of any type of human cell needed for therapeutic purposes. The table also contains interesting review articles and links to repositories through which such lines are available to researchers. This table was last updated and reviewed in June 2024.

To find additional animal models or learn more about each respective system, please examine and follow the associated literature links and references within each table.

Sources for MDF lines include:

- **The iPSC Library:** In 2016, MDF made a grant to RUCDR Infinite Biologics (DBA Sampled since 2022) and Rutgers University, which led to the creation of 21 iPSC lines from seven subjects for DM1 and DM2. These iPSC lines are available to commercial and academic users through the online catalog hosted by the National Institute of Neurological Disorders and Stroke (NINDS) Human Cell and Data Repository (NHCDR). Questions about the cell lines may be directed to Dr. Jennifer Moore, Head of Cellular Services at Sampled, at jennifer.moore@sampled.com.

- **Coriell Institute for Medical Research**

To find additional information and resources focused on myotonic dystrophy, visit the Myotonic Dystrophy Foundation website at: www.myotonic.org.

iPSC Models - Myotonic Dystrophy Animal Models & Tools

iPSC Model	Article Type
Dottori M, Li WJ, Minchiotti G, Rosa A, Sangiuolo F. Editorial: Reviews in induced pluripotent stem cells. Front Cell Dev Biol. 2023 May 4;11:1197891. doi: 10.3389/fcell.2023.1197891. PMID: 37215079; PMCID: PMC10193027.	Review
Mazaleyrat K, Badja C, Broucqsault N, Chevalier R, Laberthonnière C, Dion C, Baldasseroni L, El-Yazidi C, Thomas M, Bachelier R, Altié A, Nguyen K, Lévy N, Robin JD, Magdinier F. Multilineage Differentiation for Formation of Innervated Skeletal Muscle Fibers from Healthy and Diseased Human Pluripotent Stem Cells. Cells. 2020 Jun 23;9(6):1531. doi: 10.3390/cells9061531. PMID: 32585982; PMCID: PMC7349825.	Review
Wang Y, Wang Z, Sun H, Shi C, Yang J, Liu Y, Liu H, Zhang S, Zhang L, Xu Y, Zhang J. Generation of induced pluripotent stem cell line(ZZUi006-A)from a patient with myotonic dystrophy type 1. Stem Cell Res. 2018 Oct;32:61-64. doi: 10.1016/j.scr.2018.08.013. Epub 2018 Aug 22. PMID: 30216892.	Review
Pierre M, Jauvin D, Puymirat J, Boutjdir M, Chahine M. Generation of three myotonic dystrophy type 1 patient iPSC lines (CBRCULi018-A, CBRCULi019-A, CBRCULi020-A) derived from lymphoblastoid cell lines for disease modelling and therapeutic research. Stem Cell Res. 2024 Apr;76:103375. doi: 10.1016/j.scr.2024.103375. Epub 2024 Mar 3. PMID: 38490135.	
De Serres-Bérard T, Jauvin D, Puymirat J, Chahine M. Generation of induced pluripotent stem cell lines from pediatric patients with congenital myotonic dystrophy (CBRCULi012-A and CBRCULi013-A) and age-matched controls (CBRCULi010-A and CBRCULi011-A). Stem Cell Res. 2023 Oct;72:103234. doi: 10.1016/j.scr.2023.103234. Epub 2023 Oct 19. PMID: 37871474.	
Chahine M, Jauvin D, Pierre M, Puymirat J, Boutjdir M. Lymphoblastoid cell lines derived from iPSCs of a myotonic dystrophy type 1 patient carrying 700 CTG repeats (CBRCULi007-A) and a control (CBRCULi006-A). Stem Cell Res. 2023 Sep;71:103148. doi: 10.1016/j.scr.2023.103148. Epub 2023 Jun 17. PMID: 37352653.	
Kawada R, Jonouchi T, Kagita A, Sato M, Hotta A, Sakurai H. Establishment of quantitative and consistent in vitro skeletal muscle pathological models of myotonic dystrophy type 1 using patient-derived iPSCs. Sci Rep. 2023 Jan 11;13(1):94. doi: 10.1038/s41598-022-26614-z. PMID: 36631509; PMCID: PMC9834395.	

iPSC Model	Article Type
Herrero-Hernandez P, Bergsma AJ, Pijnappel WWMP. Generation of Human iPSC-Derived Myotubes to Investigate RNA-Based Therapies In Vitro. Methods Mol Biol. 2022;2434:235-243. doi: 10.1007/978-1-0716-2010-6_15. PMID: 35213021; PMCID: PMC9703849.	
Poulin H, Mercier A, Djemai M, Pouliot V, Deschenes I, Boutjdir M, Puymirat J, Chahine M. iPSC-derived cardiomyocytes from patients with myotonic dystrophy type 1 have abnormal ion channel functions and slower conduction velocities. Sci Rep. 2021 Jan 28;11(1):2500. doi: 10.1038/s41598-021-82007-8. PMID: 33510259; PMCID: PMC7844414.	
Dinarelli S, Girasole M, Spitalieri P, Talarico RV, Murdocca M, Botta A, Novelli G, Mango R, Sangiuolo F, Longo G. AFM nano-mechanical study of the beating profile of hiPSC-derived cardiomyocytes beating bodies WT and DM1. J Mol Recognit. 2018 Oct;31(10):e2725. doi: 10.1002/jmr.2725. Epub 2018 May 10. PMID: 29748973.	
Dastidar S, Ardui S, Singh K, Majumdar D, Nair N, Fu Y, Reyon D, Samara E, Gerli MFM, Klein AF, De Schrijver W, Tipanee J, Seneca S, Tulalamba W, Wang H, Chai YC, In't Veld P, Furling D, Tedesco FS, Vermeesch JR, Joung JK, Chuah MK, VandenDriessche T. Efficient CRISPR/Cas9-mediated editing of trinucleotide repeat expansion in myotonic dystrophy patient-derived iPS and myogenic cells. Nucleic Acids Res. 2018 Sep 19;46(16):8275-8298. doi: 10.1093/nar/gky548. PMID: 29947794; PMCID: PMC6144820.	
Spitalieri P, Talarico RV, Murdocca M, Fontana L, Marcaurelio M, Campione E, Massa R, Meola G, Serafino A, Novelli G, Sangiuolo F, Botta A. Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. Front Physiol. 2018 Jul 27;9:967. doi: 10.3389/fphys.2018.00967. PMID: 30100878; PMCID: PMC6074094.	
Mondragon-Gonzalez R, Perlingeiro RCR. Recapitulating muscle disease phenotypes with myotonic dystrophy 1 induced pluripotent stem cells: a tool for disease modeling and drug discovery. Dis Model Mech. 2018 Jul 18;11(7):dmm034728. doi: 10.1242/dmm.034728. PMID: 29898953; PMCID: PMC6078411.	
Spitalieri P, Talarico RV, Caioli S, Murdocca M, Serafino A, Girasole M, Dinarelli S, Longo G, Pucci S, Botta A, Novelli G, Zona C, Mango R, Sangiuolo F. Modelling the pathogenesis of Myotonic Dystrophy type 1 cardiac phenotype through human iPSC-derived cardiomyocytes. J Mol Cell Cardiol. 2018 May;118:95-109. doi: 10.1016/j.yjmcc.2018.03.012. Epub 2018 Mar 15. PMID: 29551391.	
Xia G, Terada N, Ashizawa T. Human iPSC Models to Study Orphan Diseases: Muscular Dystrophies. Curr Stem Cell Rep. 2018;4(4):299-309. doi: 10.1007/s40778-018-0145-5. Epub 2018 Oct 4. PMID: 30524939; PMCID: PMC6244555.	

iPSC Model	Article Type
Martineau L, Racine V, Benichou SA, Puymirat J. Lymphoblastoids cell lines - Derived iPSC line from a 26-year-old myotonic dystrophy type 1 patient carrying (CTG)200 expansion in the DMPK gene: CHUQi001-A. Stem Cell Res. 2018 Jan;26:103-106. doi: 10.1016/j.scr.2017.12.010. Epub 2017 Dec 16. PMID: 29274549.	
Martineau L, Racine V, Benichou SA, Puymirat J. Lymphoblastoids cell lines - Derived iPSC line from a 26-year-old myotonic dystrophy type 1 patient carrying (CTG)200 expansion in the DMPK gene: CHUQi001-A. Stem Cell Res. 2018 Jan;26:103-106. doi: 10.1016/j.scr.2017.12.010. Epub 2017 Dec 16. PMID: 29274549.	
Jaworska E, Kozlowska E, Switonski PM, Krzyzosiak WJ. Modeling simple repeat expansion diseases with iPSC technology. Cell Mol Life Sci. 2016 Nov;73(21):4085-100. doi: 10.1007/s00018-016-2284-0. Epub 2016 Jun 3. PMID: 27261369.	
Kalra S, Montanaro F, Denning C. Can Human Pluripotent Stem Cell-Derived Cardiomyocytes Advance Understanding of Muscular Dystrophies? J Neuromuscul Dis. 2016 Aug 30;3(3):309-332. doi: 10.3233/JND-150133. PMID: 27854224; PMCID: PMC5123622.	
Gao Y, Guo X, Santostefano K, Wang Y, Reid T, Zeng D, Terada N, Ashizawa T, Xia G. Genome Therapy of Myotonic Dystrophy Type 1 iPS Cells for Development of Autologous Stem Cell Therapy. Mol Ther. 2016 Aug;24(8):1378-87. doi: 10.1038/mt.2016.97. Epub 2016 May 12. PMID: 27203440; PMCID: PMC5023370.	
Xia G, Gao Y, Jin S, Subramony SH, Terada N, Ranum LP, Swanson MS, Ashizawa T. Genome modification leads to phenotype reversal in human myotonic dystrophy type 1 induced pluripotent stem cell-derived neural stem cells. Stem Cells. 2015 Jun;33(6):1829-38. doi: 10.1002/stem.1970. PMID: 25702800; PMCID: PMC4441571.	
Du J, Campau E, Soragni E, Jespersen C, Gottesfeld JM. Length-dependent CTG·CAG triplet-repeat expansion in myotonic dystrophy patient-derived induced pluripotent stem cells. Hum Mol Genet. 2013 Dec 20;22(25):5276-87. doi: 10.1093/hmg/ddt386. Epub 2013 Aug 9. PMID: 23933738; PMCID: PMC3842182.	
Xia G, Santostefano KE, Goodwin M, Liu J, Subramony SH, Swanson MS, Terada N, Ashizawa T. Generation of neural cells from DM1 induced pluripotent stem cells as cellular model for the study of central nervous system neuropathogenesis. Cell Reprogram. 2013 Apr;15(2):166-77. doi: 10.1089/cell.2012.0086. PMID: 23550732; PMCID: PMC3616452.	