

Cell lines

A repository of >80 **fibroblast lines** from Friedreich's ataxia patients, carriers and healthy individuals is available at UT Southwestern. The repository is a collaboration between the laboratories of Dr. Marek Napierala, at UTSW Dr. David Lynch at the Children's Hospital of Philadelphia. See the available cell lines here: <https://www.utsouthwestern.edu/labs/napierala/cell-line/> Send requests to marek.napierala@utsouthwestern.edu

Reference:

- Li Y¹ Polak U, Clark AD, Bhalla AD, Chen YY, Li J, Farmer J, Seyer L, Lynch D, Butler JS, Napierala M. (2016) Establishment and Maintenance of Primary Fibroblast Repositories for Rare Diseases- Friedreich's Ataxia Example. *Biopreserv Biobank*. 2016 Aug;14(4):324-9.

Friedreich's ataxia **lymphoblast and fibroblast** cell lines are available at the Coriell Cell Repository. Please note that several researchers have reported problems working with the fibroblast lines from Coriell (they have been passaged many times) and there are only a few lines available.

- <http://ccr.coriell.org/Sections/Search/Search.aspx?PgId=165&q=frda>.

Dr. Marek Napierala at UTSW has established a repository of Friedreich's ataxia **induced pluripotent stem cells (iPSCs)**. The repository currently includes 20 FRDA and control iPSC lines including three sets of patient and isogenic, CRISPR/Cas9-edited paired lines. See the available lines here:

<https://www.utsouthwestern.edu/labs/napierala/cell-line/> The price is \$1000 + shipping per vial but recipients of active FARA grants will only pay shipping costs. Send requests to marek.napierala@utsouthwestern.edu

FA iPSCs are also available at Coriell Cell Repository. These cell lines were established by the laboratory of Joel Gottesfeld, PhD at The Scripps Research Institute. Coriell provides a Certificate of Analysis.

- http://ccr.coriell.org/Sections/Search/Sample_Detail.aspx?PgId=166&Ref=GM23404
- http://ccr.coriell.org/Sections/Search/Sample_Detail.aspx?PgId=166&Ref=GM23913

References:

- Ku S, Soragni E, Campau E, Thomas E, Altun G, Laurent L, Loring J, Napierala M, Gottesfeld J. (2010) Friedreich's ataxia induced pluripotent stem cells model intergenerational GAA-TTC triplet repeat instability. *Cell Stem Cell*. 7:631-7 2009. (PubMed ID: 21040903)

FARA works closely with several investigators in the United States, Europe and Australia who are developing Friedreich's ataxia neuronal and cardiac cell models by differentiation of iPSCs. These models are available by collaborating with the investigators. For more information email liz.soragni@curefa.org

Other cellular models have been developed from a variety of approaches, with different defining features. Here are 2 examples of murine cell models:

- Calmels N, Schmucker S, Wattenhofer-Donzé M, Martelli A, Vaucamps N, Reutenauer L, Messaddeq N, Bouton C, Koenig M, Puccio H. (2009) The first cellular models based on frataxin

missense mutations that reproduce spontaneously the defects associated with Friedreich ataxia. *PlosOne*. **4**: e6379.

- Fil D, Chacko BK, Conley R, Ouyang X, Zhang J, Darley-Usmar VM, Zuberi AR, Lutz CM, Napierala M, Napierala JS. (2020) Mitochondrial damage and senescence phenotype of cells derived from a novel frataxin G127V point mutation mouse model of Friedreich's ataxia. *Dis Model Mech*. 2020 Jul 27;13(7):dmm045229. doi: 10.1242/dmm.045229.