Friedreich Ataxia – Omic Datasets

FARA is grateful to the research community for continued characterization and subsequent data sharing of datasets from Friedreich ataxia patient samples and animal and cell-based models.

An overview of published transcriptomic, proteomic and metabolomic studies using different FA models is shown below:

Published Transcriptomic FA Studies				
Reference	**Method**	**Sample**	**PubMed link**	**GEO database link**
Yeast				
Moreno-Cermeño A, Alsina D, Cabiscol E, Tamarit J, Ros J **(2013)** "Metabolic remodeling in frataxin-deficient yeast is mediated by Cth2 and Adr1." *Biochim Biophys Acta.* **1833***: 3*326-37.	Transcriptome profiling by microarray	Conditional yeast frataxin homologue (Yfh1) depletion mutant.	https://www.ncbi.nlm.nih.gov/pubmed/24100161	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=24100161
shRNA FXNKD Mouse Model				
Chandran V, Gao K, Swarup V, Versano R, Dong H, Jordan MC, Geschwind DH. **(2017)** "Inducible and reversible phenotypes in a novel mouse model of Friedreich's Ataxia." *Elife.* **6:** e30054.	Transcriptome profiling by microarray	Heart, cerebellum and DRG neurons from shRNA FXNKD mouse, WT, and rescue mice.	https://www.ncbi.nlm.nih.gov/pubmed/29257745	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=29257745

KIKO Mouse Model				
Coppola G, Choi S, Santos MM, Miranda CJ, Tentier D, Wexler EM, Pandolfo M, Geschwind DH. **(2006)** "Gene expression profiling in frataxin deficient mice: Microarray evidence for significant expression changes without detectable neurodegeneration." *Neurobiol Dis.* **22:**302-11.	Transcriptome profiling by microarray	3 CNS regions (spinal cord, brainstem, cerebellum).	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2886035/	
Rai M, Soragni E, Jenssen K, Burnett R, Herman D, Coppola G, Geschwind DH Gottesfeld JM, Pandolfo M. **(2008)** "HDAC inhibitors correct frataxin deficiency in a Friedreich ataxia mouse model." *PLoS One.* **3:** e1958.	Transcriptome profiling by microarray	Brain (cerebral hemispheres), cerebellum, and heart.	https://www.ncbi.nlm.nih.gov/pubmed/18463734	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=18463734
Coppola G, Marmolino D, Lu D, Wang Q, Cnop M, Rai M, Acquaviva F, Cocozza S, Pandolfo M, Geschwind DH. **(2009)** "Functional genomic analysis of frataxin deficiency reveals tissue-specific alterations and identifies the PPARgamma pathway as a therapeutic target in Friedreich's ataxia." *Hum. Mol. Genet.* **18:**2452-2461.	Transcriptome profiling by microarray	Heart, liver, and skeletal muscle.	https://www.ncbi.nlm.nih.gov/pubmed/19376812	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=19376812
Marissa Z McMackin, Blythe Durbin-Johnson, Marek Napierala, Jill S Napierala, Luis Ruiz, Eleonora Napoli, Susan Perlman, Cecilia Giulivi, Gino A Cortopassi (**2019**) Potential biomarker identification for Friedreich's ataxia using overlapping gene expression patterns in patient cells and mouse dorsal root ganglion PLoS One 14(10):e0223209	RNA-seq	DRG	https://pubmed.ncbi.nlm.nih.gov/31665133/	

Turchi R, Tortolici F, Guidobaldi G, Iacovelli F, Falconi M, Rufini S, Faraonio R, Casagrande V, Federici M, De Angelis L, Carotti S, Francesconi M, Zingariello M, Morini S, Bernardini R, Mattei M, La Rosa P, Piemonte F, Lettieri-Barbato D, Aquilano K. Frataxin deficiency induces lipid accumulation and affects thermogenesis in brown adipose tissue. Cell Death Dis. 2020 Jan 23;11(1):51	RNA-seq	Brown adipose tissue	https://pubmed.ncbi.nlm.nih.gov/31974344/	
YG8R Mouse Model				
Shan Y, Schoenfeld RA, Hayashi G, Napoli E, Akiyama T, Carstens MI, Carstens EE, Pook MA, Cortopassi GA. **(2013)** "Frataxin Deficiency Leads to Defects in Expression of Antioxidants and Nrf2 Expression in Dorsal Root Ganglia of the Friedreich's Ataxia YG8R Mouse Model. *"Antioxid Redox Signal.* **19:**1481-93.	Transcriptome profiling by microarray	DRG tissue.	https://www.ncbi.nlm.nih.gov/pubmed/23350650	
MCK-FXNKO Mouse				
Seznec H, Simon D, Bouton C, Reuenauer L, Hertzog A, Golik P, Procaccio V, Patel M, Drapier JC, Koenig M, Puccio H. **(2004)** "Friedreich ataxia: the oxidative stress paradox." *Hum. Mol. Genet.* **14:**463-74.	Mitochondrial transcriptome profiling by microarray (Mitochip, MAMMAG)	Heart of MCK-FXNKO mouse (i.e., heart and skeletal muscle FXN knockout).	https://www.ncbi.nlm.nih.gov/pubmed/15615771	

Martin AS, Abraham DM, Hershberger KA, Bhatt DP, Mao L, Cui H, Liu J, Liu X, Muehlbauer MJ, Grimsrud PA, Locasale JW, Payne RM, Hirschey MD. (**2017)** "Nicotinamide mononucleotide requires SIRT3 to improve cardiac function and bioenergetics in a Friedreich's ataxia cardiomyopathy model." *JCI Insight*. **2**: e93885.	nanoLC-MS/MS	Heart of MCK-FXNKO mouse (i.e., heart and skeletal muscle FXN knockout).	https://www.ncbi.nlm.nih.gov/pubmed/28724806	
FA Cell Culture Models; FA Fibroblasts; FA Lymphocytes; FA-Patient iPSC-Derived Cells				
Tan G, Napoli E, Taroni F, Cortopassi G. **(2003)** "Decreased expression of genes involved in sulfur amino acid metabolism in frataxin-deficient cells." *Hum. Mol. Genet.* **12:**1699-711.	Transcriptome profiling by microarray	Lymphoblasts, fibroblasts, and N-tera2/D1 (NT2) cells (with siRNA against frataxin).	https://www.ncbi.nlm.nih.gov/pubmed/12837693	
Tan G, Napoli E, Taroni F, Cortopassi G. **(2003**) "Decreased expression of genes involved in sulfur amino acid metabolism in frataxin-deficient cells." *Hum. Mol. Genet.* **12:**1699-711.	Transcriptome profiling by microarray	Lymphoblasts, fibroblasts, and N-tera2/D1 (NT2) cells (with siRNA against frataxin).	https://www.ncbi.nlm.nih.gov/pubmed/12837693	
Burnett R, Melander C, Puckett JW, Son LS, Wells RD, Dervan PB, Gottesfeld JM. **(2006)** "DNA sequence-specific polyamides alleviate transcription inhibition associated with long GAA.TTC repeats in Friedreich's ataxia." *Proc. Natl. Acad. Sci.* **103:**11497-502.	Transcriptome profiling by microarray	FA and control lymphoid cells.	https://www.ncbi.nlm.nih.gov/pubmed/16857735	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=16857735

Ku S, Soragni E, Campau E, Thomas EA, Altun G, Laurent LC, Loring JF, Napierala M, Gottesfeld JM. **(2010)** "Friedreich's ataxia induced pluripotent stem cells model intergenerational GAA·TTC triplet repeat instability." *Cell Stem Cell*. **7:**631-7.	Transcriptome profiling by microarray	FA patient fibroblast-derived iPSCs.	https://www.ncbi.nlm.nih.gov/pubmed/21040903	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=21040903
Chan PK, Torres R, Yandim C, Law PP, Khadayate S, Mauri M, Grosan C, Chapman-Rothe N, Giunti P, Pook M, Festenstein R. **(2013)** "Heterochromatinization induced by GAA-repeat hyperexpansion in Friedreich's ataxia can be reduced upon HDAC inhibition by vitamin B3." *Hum. Mol. Genet.* **22:**2662-75.	High-throughput RNA sequencing treated with nicotinamide	Patient primary lymphoblasts	https://www.ncbi.nlm.nih.gov/pubmed/23474817	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=23474817
Soragni E, Miao W, Iudicello M, Jacoby D et al. **(2014).** "Epigenetic therapy for Friedreich ataxia." *Ann Neurol* **76**(4):489-508. PMID: 25159818.	BeadArray of cells treated with HDAC109	Patient iPSC neurons	https://www.ncbi.nlm.nih.gov/pubmed/25159818	https://www.ncbi.nlm.nih.gov/gds/?Db=gds&DbFrom=pmc&Cmd=Link&LinkName=pmc_gds&IdsFromResult=4361037
Napierala JS, Li Y, Lu Y, Lin K, Hauser LA, Lynch DR, Napierala M. **(2017)** "Comprehensive analysis of gene expression patterns in Friedreich's ataxia fibroblasts by RNA sequencing reveals altered levels of protein synthesis factors and solute carriers." *Dis Model Mech.* **10**(11):1353-1369.	RNAseq	FA and control fibroblasts	https://www.ncbi.nlm.nih.gov/pubmed/29125828	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=29125828

Erwin GS, Grieshop MP, Ali A, Qi J et al. (**2017)** "Synthetic transcription elongation factors license transcription across repressive chromatin". *Science* **358**(6370):1617-1622	RNAseq of cells treated with synthetic transcription elongation factors	FA lymphocytes	https://www.ncbi.nlm.nih.gov/pubmed/29192133	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=29192133
Cotticelli MG, Xia S, Kaur A, Lin D, Wang Y, Ruff E, Tobias JW, Wilson RB. **(2018)** "Identification of p38 MAPK as a novel therapeutic target for Friedreich's ataxia." *Scientific Reports*. **8:**5007.	Transcriptome profiling by microarray	FA fibroblast lines (growth defect and cytokine secretion defect).	https://www.ncbi.nlm.nih.gov/pubmed/29257745	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=29257745
Lai JI, Nachun D, Petrosyan L, Throesch B, Campau E, Gao F, Baldwin KK, Coppola G, Gottesfeld JM, Soragni E. (2019) Transcriptional profiling of isogenic Friedreich ataxia neurons and effect of an HDAC inhibitor on disease signatures. J Biol Chem. 2019 Feb 8;294(6):1846-1859.	RNA-seq	iPSC-derived neurons (CNS and sensory)	https://pubmed.ncbi.nlm.nih.gov/30552117/	https://www.ncbi.nlm.nih.gov/bioproject/PRJNA495860/
Tiano F, Amati F, Cherubini F, Morini E, Vancheri C, Maletta S, Fortuni S, Serio D, Quatrana A, Luffarelli R, Benini M, Alfedi G, Panarello L, Rufini A, Toschi N, Frontali M, Romano S, Marcotulli C, Casali C, Gioiosa S, Mariotti C, Mongelli A, Fichera M, Condò I, Novelli G, Testi R, Malisan F. Frataxin deficiency in Friedreich's ataxia is associated with reduced levels of HAX-1, a regulator of cardiomyocyte death and survival. Hum Mol Genet. 2020 Feb 1;29(3):471-482	Transcriptome profiling by microarray	lymphoblastoid cells	https://pubmed.ncbi.nlm.nih.gov/31943004/	

FA Patient Peripheral Blood				
Haugen AC, Prospero NA, Parker JS, Fannin RD, Chou J, Meyer JN, Halweg C, Collins JB, Durr A, Fischbeck K, Van Houten B. **(2010)** "Altered gene expression and DNA damage in peripheral blood cells from Friedreich's ataxia patients: cellular model of pathology." *PLoS One*. **6**: e1000812.	Transcriptome profiling by microarray	Peripheral blood.	https://www.ncbi.nlm.nih.gov/pubmed/20090835	
Coppola G, Burnett R, Perlman S, Versano R, Gao F, Pasterer H, Rai M, Sacca F, Filla A, Lynch DR, Rusche JR, Gottesfeld JM, Pandolfo M, Geschwind DH**. (2011)** "A gene expression phenotype in lymphocytes from Friedreich ataxia patients." *Ann Neurol.* **70**:790-804.	Transcriptome profiling by microarray	Peripheral blood mononuclear cells.	https://www.ncbi.nlm.nih.gov/pubmed/22162061	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=22162061
Salehi MH, Kamalidehghan B, Houshmand M, Yong Meng G, Sadeghizadeh M, Aryani O, et al. **(2014)** "Gene Expression Profiling of Mitochondrial Oxidative Phosphorylation (OXPHOS) Complex I in Friedreich Ataxia (FRDA) Patients." *PLoS ONE.* **9:** e94069."	Gene expression profiling (RT-PCR) of mitochondrial and nuclear DNA genes that encode for OXPHOS.	Blood.	https://www.ncbi.nlm.nih.gov/pubmed/24705504	
Steinkellner H, Singh HN, Mcukenthaler MU, Golderberg H, Moganty RR, Scheiber-Mojdehkar B, Sturm B. **(2017)** "No changes in heme synthesis in human Friedreich´s ataxia erythroid progenitor cells." *Gene*. **621:**5-11.	"cDNA-microarray analysis (“IronChip”; the Chip measures the expression of 536 genes with a special focus on genes which are involved in iron/copper metabolism)."	Erythroid progenitor cells.	https://www.ncbi.nlm.nih.gov/pubmed/28412459	

Seco-Cervera M González-Rodríguez D, Ibáñez-Cabellos JS, Peiró-Chova L, González-Cabo P, García-López E, Vílchez JJ, Sanz-Gallego I, Pallardó FV García-Giménez JL **(2017)** "Circulating miR-323-3p is a biomarker for cardiomyopathy and an indicator of phenotypic variability in Friedreich's ataxia patients". *Sci Rep.* **7**(1):5237.	RNAseq for miRNA	Plasma	https://www.ncbi.nlm.nih.gov/pubmed/28701783	
Dantham S, Srivastava AK, Gulati S, Rajeswari MR. **(2018)** "Differentially Regulated Cell-Free MicroRNAs in the Plasma of Friedreich's Ataxia Patients and Their Association with Disease Pathology". *Neuropediatrics.* **49**(1):35-43.	Agilent microarray platform for miRNA	Plasma	https://www.ncbi.nlm.nih.gov/pubmed/29179232	
Nachun D, Gao F, Isaacs C, Strawser C, Yang Z, Dokuru D, Van Berlo V, Sears R, Farmer J, Perlman S, Lynch DR, Coppola G**. (2018)** "Peripheral blood gene expression reveals an inflammatory transcriptomic signature in Friedreich's ataxia patients." *Hum. Mol. Genet.* **ddy198**. *	Transcriptome profiling by microarray	Peripheral blood.	https://www.ncbi.nlm.nih.gov/m/pubmed/29790959/	

* FARA is working with Giovanni Coppola, PhD at UCLA and the Collaborative Clinical Research Network (CCRN) and others to develop a database of gene expression data from Friedreich's ataxia patients and from mouse models of the disease. The database is available from Dr. Coppola's website at https://coppolalab.ucla.edu/account/login/ . You will need to register for a password to access the database. Array data is available for some patients who have taken part in natural history studies through the CCRN, so clinical data can be accessed. Additional data is still being added over time, and additional analyses are underway. For more information, please contact Giovanni Coppola: gcoppola@ucla.edu

Published Large Scale Methylation FA Studies				
Reference	**Method**	**Sample**	**PubMed link**	**GEO database link**
FA-Patient iPSC-Derived Cells				
Evans-Galea MV, Carrodus N, Rowley SM, Corben LA, Tai G, Saffery R, Galati JC, Wong NC, Craig JM, Lynch DR, Regner SR, Brocht AF, Perlman SL, Bushara KO, Gomez CM, Wilmot GR, Li L, Varley E, Delatycki MB, Sarsero JP **(2012)** "FXN methylation predicts expression and clinical outcome in Friedreich ataxia.". *Ann Neurol.* **71**(4):487-97.	EpiTYPER MassARRAY platform to determine C methylation upstream and downstream of the GAA	FA Lymphocytes	https://www.ncbi.nlm.nih.gov/pubmed/22522441	
Rodden LN, Chutake YK, Gilliam K, Lam C, Soragni E, Hauser L, Gilliam M, Wiley G, Anderson MP, Gottesfeld JM, Lynch DR, Bidichandani SI. Methylated and unmethylated epialleles support variegated epigenetic silencing in Friedreich ataxia. Hum Mol Genet. 2021 Feb 4;29(23):3818-3829	Bisulfite Deep sequencing	PBMCs, iPSC-derived neurons	https://pubmed.ncbi.nlm.nih.gov/33432325/	

Published Proteomic FA Studies				
Reference	**Method**	**Sample**	**PubMed link**	**GEO database link**
Yeast				
Alsina D, Ros J, and Tamarit J. **(2018)** "Nitric oxide prevents Aft1 activation and metabolic remodeling in frataxin-deficient yeast." *Redox Biol.* **14***:* 131-41.	Proteomics (MRM: Multiple Reaction Monitoring; targeted mass spectrometry)	Conditional yeast frataxin homologue (Yfh1) depletion mutant.	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5602528/	
Moreno-Cermeño A, Alsina D, Cabiscol E, Tamarit J, Ros J **(2013)** "Metabolic remodeling in frataxin-deficient yeast is mediated by Cth2 and Adr1." *Biochim Biophys Acta.* **1833***:* *3*326-37.	Proteomics (2D-electrophoresis with tryptic digestion and MS/MS ion search)	Conditional yeast frataxin homologue (Yfh1) depletion mutant.	https://www.ncbi.nlm.nih.gov/pubmed/24100161	https://www.ncbi.nlm.nih.gov/gds?LinkName=pubmed_gds&from_uid=24100161
Irazusta V, Cabiscol E Reverter-Branchat G, Ros J, Tamarit J. **(2006)** "Manganese is the link between frataxin and iron-sulfur deficiency in the yeast model of Friedreich ataxia." *J Biol Chem.* **281:**12227-32.	Proteomics (2D-electrophoresis)	Conditional yeast frataxin homologue (Yfh1) depletion mutant.	https://www.ncbi.nlm.nih.gov/pubmed/16510442	
Gabrielli N, Ayté J, Hidalgo E. **(2012)** "Cells Lacking Pfh1, a Fission Yeast Homolog of Mammalian Frataxin Protein, Display	Dimethyl-labeled proteins used for MS/MS	Conditional yeast pfh1, coding for the fission yeast homolog	https://www.ncbi.nlm.nih.gov/pubmed/23115244	

Constitutive Activation of the Iron Starvation Response" *J Biol Chem*. **287**:43042-51.		of human frataxin, depletion mutant.		
MCK-FXNKO Mouse				
Sutak R, Xu X, Whitnall M, Kashem MA, Vyoral D, Richardson DR. **(2008)** "Proteomic analysis of hearts from frataxin knockout mice: marked rearrangement of energy metabolism, a response to cellular stress and altered expression of proteins involved in cell structure, motility and metabolism." *Proteomics*. **8:** 1731-41.	Proteomics (2D-electrophoresis with tryptic digestion and MS/MS ion search)	Heart of MCK-FXNKO mouse (i.e., heart and skeletal muscle FXN knockout).	https://www.ncbi.nlm.nih.gov/pubmed/18340635	
Martin AS, Abraham DM, Hershberger KA, Bhatt DP, Mao L, Cui H, Liu J, Liu X, Muehlbauer MJ, Grimsrud PA, Locasale JW, Payne RM, Hirschey MD. **(2017)** "Nicotinamide mononucleotide requires SIRT3 to improve cardiac function and bioenergetics in a Friedreich's ataxia cardiomyopathy model." *JCI Insight*. **2**: e93885.	nanoLC-MS/MS	Heart of MCK-FXNKO mouse (i.e., heart and skeletal muscle FXN knockout).	https://www.ncbi.nlm.nih.gov/pubmed/28724806	
FA-Patient iPSC-Derived Cells				
Shan B, Xu C, Zhang Y, Xu T, Gottesfeld JM, and Yates JB 3rd**. (2014)** "Quantitative proteomic analysis identifies targets and pathways of a 2-aminobenzamide HDAC inhibitor in Friedreich's ataxia patient iPSC-derived neural stem cells." *J Proteome Res.* **13***:* 4558-66.	Proteomics (MuDPIT: Multi-Dimensional Protein Identification Technology)	FA-patient derived iPSC neural stem cell; Nuclear extraction.	https://www.ncbi.nlm.nih.gov/pubmed/24933366	
FA Patient Lymphocytes/fiborblasts				

Télot L, Roussseau E, Lesuisse E, Garcia C, Morlet B, Léger T, Camadro JM, and Serre V. **(2018)** "Quantitative proteomics in Friedreich's ataxia B-lymphocytes: A valuable approach to decipher the biochemical events responsible for pathogenesis." *Biochim Biophys Acta.* **1864***:* 997-1009.	LC-MS/MS quantitative proteomics	FA B-lymphoblasts.	https://www.ncbi.nlm.nih.gov/pubmed/29329987	
Pathak D, Srivastava AK, Padma MV, Gulati S, Rajeswari MR. Quantitative Proteomic and Network Analysis of Differentially Expressed Proteins in PBMC of Friedreich's Ataxia (FRDA) Patients. Front Neurosci. 2019 Oct 14;13:1054.	2D-Differential in-Gel Electrophoresis	PBMCs	https://pubmed.ncbi.nlm.nih.gov/31680804/	
Napierala JS, Rajapakshe K, Clark A, Chen YY, Huang S, Mesaros C, Xu P, Blair IA, Hauser LA, Farmer J, Lynch DR, Edwards DP, Coarfa C, Napierala M. Reverse phase protein array reveals correlation of retinoic acid metabolism with cardiomyopathy in Friedreich's ataxia. Mol Cell Proteomics. 2021 May 12:100094. doi: 10.1016/j.mcpro.2021.100094	Reverse phase protein array	Fibroblasts	https://pubmed.ncbi.nlm.nih.gov/33991687/	
Patient Plasma				
Swarup V, Srivastava AK, Padma MV, Rajeswari MR. **(2013)** "Quantitative profiling and identification of differentially expressed plasma proteins in Friedreich's ataxia." *J Neurosci Res*. **91**: 1483-91.	Proteomics (2D-electrophoresis of protein samples labeled with fluorescent cyanine (Cy) dyes; internal standard prepared by mixing equal amounts of all	Patient plasma.	https://www.ncbi.nlm.nih.gov/pubmed/23996585	

	samples with a Cy dye). LC-MS/MS			
Bulteau AL, Planamente S, Jornea L, Dur A, Lesuisse E, Camadro JM, Auchere F. **(2012)** "Changes in mitochondrial glutathione levels and protein thiol oxidation in Yfh1 deficient yeast cells and the lymphoblasts of patients with Friedreich's ataxia." *Biochim Biophys Acta*. **1822**: 212-25.	Proteomic analysis of glutathionylated mitochondrial proteins (MALDI MS and MS/MS)	Patient plasma.	https://www.ncbi.nlm.nih.gov/pubmed/22200491	

Published Metabolomic FA Studies				
Reference	**Method**	**Sample**	**PubMed link**	
KIKO FA Mouse Model				
Hayashi G, Shen Y, Pedersen TL, Newman JW, Pook M, Cortopassi G. **(2014)** "Frataxin deficiency increases cyclooxygenase 2 and prostaglandins in cell and animal models of Friedreich's ataxia." *Hum. Mol. Genet.* **23:**6838-47.	UPLC-tandem mass spectrometry	KIKO mouse cerebellum.	https://www.ncbi.nlm.nih.gov/pubmed/ 25104852	
MCK-FXNKO Mouse				
Martin AS, Abraham DM, Hershberger KA, Bhatt DP, Mao L, Cui H, Liu J, Liu X, Muehlbauer MJ, Grimsrud PA, Locasale JW, Payne RM, Hirschey MD. **(2017)** "Nicotinamide mononucleotide requires SIRT3	nanoLC-MS/MS	Heart of MCK-FXNKO mouse (i.e., heart and skeletal	https://www.ncbi.nlm.nih.gov/pubmed/28724806	

to improve cardiac function and bioenergetics in a Friedreich's ataxia cardiomyopathy model." *JCI Insight*. **2**: e93885.		muscle FXN knockout).		
FA Patient Platelets				
Worth AJ, Basu SS, Deutsch EC, Hwang WT, Snyder NW, Lynch DR, Blair IA. **(2015)** "Stable isotopes and LC–MS for monitoring metabolic disturbances in Friedreich's ataxia platelets." *Bioanalysis*. **7:**1843-55.	Stable isotope labeling; LC-MS	Patient platelets.	https://www.ncbi.nlm.nih.gov/pubmed/26295986	