

**United Mitochondrial Disease Foundation
Research Grant Program
Projects Funded Through 2010**

PROJECT TITLE	AMOUNT AWARDED	LENGTH (YRS.)	INVESTIGATOR(S)	AFFILIATION	YEAR
Spinocerebellar ataxia with axonal neuropathy: defining the mitochondrial component.	\$130,348		Cornelius Franciscus Boerkoel, M.D., Ph.D.	Department of Medical Genetics, University of British Columbia	2010
DMCA and Barth Syndromes- similar diseases caused by defects in mitochondrial protein import?	\$110,000		Robert E. Jensen, Ph.D.	Department of Cell Biology, Johns Hopkins University	2010
Pilot study to investigate the efficacy of L-arginine therapy on endothelium-dependent vasodilation & mitochondrial metabolism in MELAS syndrome.	\$75,000		Ingrid Tein, M.D.	Division of Neurology, Hospital for Sick Children, Toronto, Canada	2010
Mitochondrial dysfunction, exercise intolerance and myopathy in skeletal muscle-specific PGC-1 α -deficient mice.	\$130,000		Christoph Handschin	University of Basel, Switzerland	2009
Development of a Novel Mass Spectrometric Approach to Measure Mitochondrial Oxidative Damage <i>In Vivo</i> .	\$110,000		Michael P. Murphy	Medical Research Council, Dunn Human Nutrition Unit, Cambridge, U.K.	2009
Selecting for Transformation with Mitochondrial DNA	\$81,857		Patrick H. O'Farrell	University of California-San Francisco	2009
Investigating the Rescue of Mitochondrial Dysfunction by SIRT1 and Calorie Restriction.	\$90,804		Sarika Srivastava	Harvard Medical School	2009
Evaluation of novel zinc-finger nucleases as a means to target m.3243 A>G in vivo.	\$99,998	2	Sion Williams (mentor Carlos Moraes)	University of Miami	2008
The role of the PINK1/Parkin Pathway in mitochondrial integrity	\$83,334	2	Leo Pallanck	University of Washington	2008
Cerium Oxide Nanoparticles in the Treatment of Mitochondrial Diseases	\$100,469	1	Beverly Rzigalinski	Virginia College of Osteopathic Medicine	2008

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Evaluation of the efficacy and safety of erythrocyte encapsulated thymidine phosphorylase therapy in two patients with Mitochondrial neurogastrointestinal encephalomyopathy	\$116,428	2	Bridget Bax	St. George's University of London	2008
Utilization of knockout mouse models to elucidate the importance of the de novo mitochondrial fatty acid synthesis pathway in mitochondrial function	\$126,563	2	Stuart Smith	Children's Hospital & Research Center at Oakland	2008
Selective alteration of mitochondrial gene expression via modulation of the dual-function h-mtTFB1 and B2 factors as a potential therapy for mitochondrial diseases	\$99,998	2	Timothy Shutt (mentor Gerald Shadel)	Yale University School of Medicine	2008
Mitochondrial Fusion Defects in Neurological Disease	\$98,300	2	Elizabeth Amriott (Mentor-Janet Shaw)	University of Utah	2008
Oxphos modulation by mitochondrial protein phosphorylation in mtDNA mutant cells	\$99,990	2	Rebeca Acin-Perez (Mentor-Giovanni Manfredi)	Weill Medical College, Cornell University	2008
Identifying genetic modifiers of tissue-specific mitochondrial DNA segregation.	\$100,000	2	Brendan Battersby	Biomedicum Helsinki, University of Helsinki	2008
Study of redox regulated pathways in the mitochondrion	\$100,000	2	Deepa Dabir (mentor Carla Koehler)	University of California - Los Angeles	2008
Mitochondrial calcium signaling and organelle dysfunction in mitochondrial diseases: molecular determinants and regulatory mechanisms	\$86,250	2	Pinton, Paolo, PhD	University of Ferrara, Italy	2007
Mitochondrial DNA synthesis and Krebs (tricarboxylic acid) cycle: the succinyl-CoA synthase	\$98,340	2	Saada (Reisch), Ann, PhD	Hadassah Hebrew University Medical Center	2007
Mutant Complex I in Drosophila melanogaster: a Novel Genetic Model for Mitochondrial Disease	\$111,779	2	Graham, Brett, PhD	Baylor College of Medicine	2007

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Genotype-Phenotype Correlation and Genetic Modifiers in Barth Syndrome	\$157,450	2	Ren, Mindong, PhD	New York University School of Medicine	2007
Increased Mitochondrial Biogenesis as Therapy to Mitochondrial Myopathies	\$94,481	2	Wenz, Tina, PhD	University of Miami	2007
Defining copper homeostasis in the mitochondria: Recruitment and distribution of copper for the assembly of cuproenzymes.	\$99,000	2	Cobine, Paul, PhD	University of Utah	2007
Development of high throughput mtDNA sequencing for mutation detection and heteroplasmy assessment.	\$110,000	2	Khrapko, Konstantin, PhD	Beth Israel Deaconess Med Ctr/Harvard Med School	2007
Determination of the nuclear transcriptional responses that affect animal physiopathology upon impaired mitochondrial respiratory chain function	\$100,000	2	Walter, Ludivine, PhD	Cornell University	2007
Development of high throughput assays for mitochondrial respiratory chain function	\$118,648	2	King, Michael, PhD	Thomas Jefferson University	2007
Molecular genetic dissection of mitochondrial complex I assembly	\$114,189	2	Hamel, Patrice, PhD	Ohio State University	2007
Identification of novel genes associated with isolated complex I deficiency using whole genome mapping in small consanguineous families	\$60,500	1	Elpeleg, Orly, MD	Hadassah Hebrew University Medical Center	2007
Utilization of fission yeast as a model for mitochondrial morphology: a new approach to discover novel genes involved in animal cells	\$98,500	2	Chiron, Stephanie, PhD	Univ of California, San Diego	2006
Molecular Signatures of Mitochondrial Disorders	\$88,850	2	Gohil, Vishal, PhD	Massachusetts General Hospital, Harvard Medical School, Boston, MA	2006
Animal models of human Barth Syndrome, a mitochondrial cardiolipin disorder	\$110,000	2	Khuchua, Zaza, PhD	Vanderbilt University Medical Center, Nashville, TN	2006

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Enzymatic, Assembly and Genetic Studies on the Human Pyruvate Dehydrogenase Multienzyme Complex	\$43,494	1	Lindsay, John, PhD	University of Glasgow, UK	2006
Enzyme Replacement Therapy: A novel Approach for Treating A Mitochondrial Disease - LAD Deficiency	\$115,000	2	Lorberboum-Galski, Haya, PhD	Hebrew University of Jerusalem, Israel	2006
Mitochondrial Ribosomal Proteins: Candidate Genes for Mitochondrial Disease	\$125,000	2	O'Brien, Thomas, PhD	University of Florida, Gainesville, FL	2006
Developing Therapies for Mitochondrial Disease	\$98,457	2	Palladino, Michael, PhD	Univ of Pittsburgh Medical School, PA	2006
Defective biogenesis of mitochondrial beta-barrel proteins as a cause for Mohr-Tranebjaerg syndrome	\$98,000	2	Rapaport, Doron, PhD	Institute for Physiological Chemistry, Munich, Germany	2006
High throughput screening for mitochondrial enhancers	\$125,000	2	Robinson, Brian, PhD	Hospital for Sick Children, Toronto, Canada	2006
Mechanisms of muscle dysfunction studied in mouse models of mitochondrial myopathies	\$122,720	2	Westerblad, Hakan, MD, PhD	Karolinska Institutet, Stockholm, Sweden	2006
Biochemical and structural studies on mitochondrial disease mutations in methionyl-tRNA	\$110,980	2	Spremulli, Linda, PhD	Univ of NC, Chapel Hill	2005
The assembly pathway of human cytochrome-c oxidase studied with RNA interference	\$86,455	1	Taanman, Jan-Willem, PhD	Royal Free & University College Medical School, University College London, UK	2005
The population prevalence of ten mtDNA mutations	\$162,878	3	Chinnery, Patrick, PhD	University of Newcastle upon Tyne	2005
The mechanism of mitochondrial dysfunction in paraplegin-deficient mice	\$126,500	3	Rugarli, Elena, MD	Telethon Institute of Genetics and Medicine, Naples, Italy	2005
Role of mitofusin-2, a mitochondria-shaping protein mutated in Charcot-Marie-Tooth 2a, in controlling mitochondrial function and apoptosis	\$94,000	2	Scorrano, Luca, MD, PhD	Dulbecco-Telethon Institute, Padova, Italy	2005

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MitoPLD, novel enzymatic regulator of mitochondrial morphology and fusion	\$141,027	2	Frohman, Michael, MD, PhD	Stony Brook University, Stony Brook, NY	2005
Barth Syndrome: A mitochondrial disease with insights into cardiolipin synthesis	\$70,525	2	Lewin, Tal, Mia, PhD	University of North Carolina at Chapel Hill	2005
Molecular Basis of Mitochondrial Gene Regulation	\$116,133	2	Churchill, Mair, PhD	University of Colorado Health Sciences Center	2005
Diagnostic Utility of DHPLC in Mitochondrial Disease	\$109,991	2	Haas, Richard, MB, B.Chir.	University of CA San Diego	2005
Understanding the role of mitochondrial fusion in mitochondrial myopathies	\$128,000	1	David Chan, MD, PhD	California Institute of Technology, Division of Biology, Pasadena, CA	2004
The nuclear-encoded gene OMI and mitochondrial disease	\$108,305	1	Miriam Meisler, PhD	University of Michigan, Dept. of Human Genetics, Ann Arbor, MI	2004
Development of a method for transforming mitochondria in living mammalian cells with exogenous DNA	\$99,360	1	Volkmar Weissig, PhD, ScD	Northeastern University, Dept. of Pharmaceutical Sciences, Boston, MA	2004
Genomic Approaches to Human Cytochrome c Oxidase Deficiency	\$90,200	1	Vamsi Mootha, MD	Massachusetts Institute of Technology, Broad Institute, Cambridge, MA	2004
The hypoxia sensing transcription factor EPAS1/HIF-2a is a novel mitochondrial disease candidate in mice and man	\$88,852	1	Joseph Garcia, MD, PhD	University of Texas SW Medical Center at Dallas, Dept. of Internal Medicine, Dallas, TX	2004
Protein phosphatase 2A in mitochondrial function and disease	\$88,000	1	Stefan Strack, PhD	University of Iowa, Carver College of Medicine, Dept. of Pharmacology, Iowa City, IA	2004
Drug development for the regulation of respiratory chain components in mitochondria	\$44,000	1	Brian Robinson, PhD	Hospital for Sick Children, Metabolism Research Programme, Toronto, Canada	2004

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GSH levels, reactive oxygen species production, lipid peroxidation products and mitochondrial membrane potential in patients with mitochondrial disease	\$34,179	1	Gregory Enns, MB, ChB	Stanford University, Dept. of Pediatrics, Stanford, CA	2004
Restoration of thymidine phosphorylase activity in MNGIE patients through platelets infusion	\$33,776	1	Ramon Marti, PhD	Fundacio Institut Hospital Universitari Vall d'Hebron, Dept. CIBBIM, Barcelona, Spain	2004
Application of RNA interference in the study of NADH-ubiquinone oxidoreductase (complex I) assembly in mammalian mitochondria	\$100,000	1	Immo Scheffler, PhD	University of California, San Diego, La Jolla, CA	2003
Selective Elimination of Defective Mitochondrial Genomes as an Approach to the Reversal of NARP and MILS Syndromes, Heritable Mitochondrial Disorders	\$100,000	1	Mikhail Alexeyev, PhD	University of South Alabama, Mobile, AL	2003
Role of Rhomboid Proteolysis in Optic Atrophy	\$90,000	1	Matthew Freeman, PhD	Laboratory of Molecular Biology – M.R.C., Cambridge, UK	2003
Molecular Basis of Mitochondrial Membrane Dynamics: a New Paradigm of Human Disease	\$83,400	1	Koji Okamoto, PhD	University of Utah, Salt Lake City, UT	2003
The Use of the Yeast CYB2 Gene As Therapy for Complex I Mutations in a C. elegans Model System	\$76,780	1	Bernard Lemier, PhD	University of Alberta, Edmonton, AB, Canada	2003
MtDNA complementation and recombination in mitochondrial disorders	\$50,000	1	Giovanni Manfredi, MD, PhD	Weill Medical College of Cornell University, New York, NY	2003
Electrophysiologic Properties of Neural Stem Cells from Patients with Mitochondrial Disease	\$81,574	1	Philip Schwartz, PhD	Children's Hospital of Orange County, Orange, CA	2002

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Exploiting the potential of yeast NDI1 gene in the therapy of diseases linked with mtDNA mutations	\$66,000	1	Yidong Bai, PhD	University of Texas Health Science Center at San Antonio, TX	2002
Exercise-induced mitochondrial gene shifting: Resistance training as a therapy for sporadic mtDNA mutations	\$61,389	1	Tanja Taivassalo, PhD	Institute for Exercise & Environmental Medicine, Dallas, TX	2002
Transport diseases in mitochondria: Full screening of DNA alterations in human genes encoding TOMM and TIMM complexes in patients with mitochondrial diseases	\$41,085	1	Jose Hernandez-Yago, PhD	Institute for Cell Research, Valencia, Spain	2002
Biochemical Basis for Maternally Inherited Deafness	\$33,000	1	Min-Xin Guan, PhD	Cincinnati Children's Hospital Medical Center, Cincinnati, OH	2001
Efficacy of prenatal diagnosis of mitochondrial diseases	\$15,000	1	Brian Robinson, PhD	The Hospital for Sick Children, Toronto, ON, Canada	2001
Complex I: The role of nuclear genes in disorders of childhood due to mitochondrial Complex I deficiency	\$12,000	1	Edwin Kirk, MD	Sydney Children's Hospital, Randwick, NSW, Australia	2001
Quantitative In Vivo 1H Magnetic Resonance Spectroscopic Imaging of Cerebral Lactate as a Screening Test for Mitochondrial Disorders	\$36,719	1	Dikoma Shungu, PhD	Columbia University, New York, NY	2000
Is oxidative damage a result of metabolic abnormalities in Alzheimer disease?	\$18,281	1	George Perry, PhD	Case Western Reserve University, Cleveland, OH	2000
Characterization of Mitochondrial Nitric-Oxide Synthase	\$31,137	1	Cecilia Giulivi, PhD	University of Minnesota, Duluth, MN	1999
Efficacy of Prenatal Diagnosis of Mitochondrial Diseases	\$8,863	1	Brian Robinson, PhD	The Hospital for Sick Children, Toronto, ON, Canada	1999
Gene mutations in Leigh's Disease	\$30,000	1	John Shoffner, MD	Horizon Molecular Laboratory, Norcross, GA	1998
Mitochondrial etiologies of pseudo-obstruction and dysmotility in children	\$5,000	1	Carolyn Bay, MD	Children's Hospital of Pittsburgh, Pittsburgh, PA	1998

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Search for Pathogenic Mitochondrial DNA Mutations Using Temporal Temperature Gradient Gel Electrophoresis (TTGE)	\$30,000	1	Richard Boles, MD	Children's Hospital of Los Angeles, Los Angeles, CA	1997
Total:	\$6,472,136				