

# Mitochondrial Disease Common Data Element (CDE) Working Group Roster

## Previous Oversight Committee Members

Danuta Krotoski, PhD - National Institutes of Health, Bethesda, Maryland (2018-2021)	
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## Biomarkers Working Group

**John Shoffner, MD - Atlanta, Georgia, Chair**

Mark Bamberger, PhD - Stealth Peptides, Inc, Newton, Massachusetts	Anne Lombes, MD, PhD - INSERM, Paris, France
Yasutoshi Koga, MD, PhD - Kurume University, Fukuoka-ken, Japan	Fernando Scaglia, MD - Baylor College of Medicine, Houston, Texas

## Cognitive/Behavioral/Psychological Outcomes Working Group

**Lisa Emrick, MD - Baylor College of Medicine, Houston, Texas, Chair**

Rebecca Vaurio, PhD - Kennedy Krieger Institute, Baltimore, Maryland	
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## Endocrinology/Diabetes/GI/Nutrition Working Group

**Shana McCormack, MD - Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, Chair**

Jose E. Abdenur, MD - Children's Hospital of Orange County, Orange, California	Philip Morgan, MD - Mitochondrial Research Guild, Seattle, Washington
Bret Goodpaster, PhD - Sanford Burnham Prebys Medical Discovery Institute, Orlando, Florida	Fernando Scaglia, MD - Baylor College of Medicine, Houston, Texas
Robert O. Heuckeroth, MD, PhD - Children's Hospital of Philadelphia, Philadelphia, Pennsylvania	Charles A. Stanley, MD - Children's Hospital of Philadelphia, Philadelphia, Pennsylvania
Robert McFarland, DoH, HEFCE - Newcastle University, Newcastle upon Tyne, United Kingdom	Steven M. Willi, MD - Children's Hospital of Philadelphia, Philadelphia, Pennsylvania

## Exercise Physiology Working Group

**Tanja Taivassalo, PhD - McGill University, Montreal, Canada, Chair**

Amy Goldstein, MD - Children's Hospital of Pittsburgh, Pittsburgh, Pennsylvania	Mark Tarnopolsky, MD, PhD - McMaster University, Hamilton, Ontario, Canada
Ronald Haller, MD - Southwestern Medical Center, Dallas, Texas	Karim Wahbi, MD, PhD - Institute of Myology, Paris, France

*Please note that the institutions for all individuals acknowledged in this section were those they belonged to when they joined the Working Group*

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## Genetics Working Group

**David Dimmock, MD - Medical College of Wisconsin, Milwaukee, Wisconsin, Co-Chair**

**Marni J. Falk, MD - Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, Co-Chair**

Xiaowu Gai, PhD - Ocular Genomics Institute, Boston, Massachusetts	Fernando Scaglia, MD - Baylor College of Medicine, Houston, Texas
Michio Hirano, MD - Columbia University Medical Center, New York, New York	Curt Scharfe, MD, PhD - Stanford University School of Medicine, Stanford, California
Anne Lombes, MD, PhD - INSERM, Paris, France	David Thorburn, PhD, FHGSA, FFSC (RCPA) - Murdoch Children's Research Institute, Victoria, Australia
Shamima Rahman, PhD, FRCP(C), FRCPC - UCL Institute of Child Health, London, United Kingdom	Stephan Zuchner, MD, PhD - Dr. John T. Macdonald Foundation, Miami, Florida

## Imaging Working Group

**David M. Mirsky, MD - Children's Hospital Colorado, Aurora, Colorado, Chair**

Abigail Collins, MD - Children's Hospital Colorado, Aurora, Colorado	Edward Yang, MD - Boston Children's Hospital, Boston, Massachusetts
Andrea L. Gropman, MD - Children's National Medical Center, Washington, District of Columbia	

## Neurological Assessments Working Group

**Bruce Cohen, MD, FAAN - Akron Children's Hospital, Akron, Ohio, Co-Chair**

**Ingrid Tein, MD, FRCP - University of Toronto, Toronto, Canada, Co-Chair**

Laurence Bindoff, MD, PhD - University of Bergen, Bergen, Norway	Saskia Koene, MD - Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands
Michio Hirano, MD - Columbia University Medical Center, New York, New York	Pascal Laforet, MD, PhD - Assistance Publique-Hopitaux de Paris, Paris, France
Matthew Klein, MD, MS, FACS - Edison Pharmaceuticals, Mountain View, California	Margherita Milone, MD, PhD - Mayo School of Graduate Medical Education, Rochester, Minnesota
Thomas Klopstock, MD - Ludwig Maximilian University of Munich, Munchen, Germany	Jan Smeitink, MD, PhD - Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands

## Patient Reported Outcomes/QOL Working Group

**Mary Kay Koenig, MD - University of Texas Health Science Center, Houston, Texas, Co-Chair**

**Sumit Parikh, MD - Cleveland Clinic, Cleveland, Ohio, Co-Chair**

Cristy Balcells, RN, MSN - Mito Action, Medford, Massachusetts	Russel Saneto, DO, PhD - Seattle Children's Hospital, Seattle, Washington
Michio Hirano, MD - Columbia University Medical Center, New York, New York	Peter W. Stacpoole, MD, PhD - University of Florida, Gainesville, Florida
Amel Karaa, MD - Massachusetts General Hospital, Boston, Massachusetts	Philip Yeske, PhD - United Mitochondrial Disease Foundation, Pittsburgh, Pennsylvania
Robert McFarland, DoH, HEFCE - Newcastle University, Newcastle upon Tyne, United Kingdom	

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## Vision Working Group

**Patrick Yu-Wai-Man, MBBS, PhD, FRCOphth - Newcastle University, Newcastle upon Tyne, United Kingdom, Chair**

Piero Barboni, MD - University of Bologna, Bologna, Italy	Tony Moore, MA, FRCS, FCROphth, FMedSci - Moorfields Eye Hospital, London, United Kingdom
Valerio Carelli, MD, PhD - University of Bologna School of Medicine, Bologna, Italy	Alfredo A. Sadun, MD, PhD - University of California Los Angeles, Los Angeles, California
Patrick Chinnery, PhD, MRCPATH, MRCP - Newcastle University, Newcastle upon Tyne, United Kingdom	Claire Sheldon, MD, PhD - University of Pennsylvania, Philadelphia, Pennsylvania
Graham E. Holder, PhD - Moorfields Eye Hospital, London, United Kingdom	Marcela Votruba, PhD, FRCOphth - Cardiff University, Cardiff, United Kingdom
John L. Keltner, MD - UC Davis School of Medicine, Sacramento, California	

## NIH Coordinating Team

Vernon Anderson, PhD - National Institute of General Medical Sciences (NIGMS), National Institutes of Health (NIH), Bethesda, Maryland	Petra Kaufmann, MD, MSc - National Institute of Neurological Disorders and Stroke (NINDS), National Institutes of Health (NIH), Bethesda, Maryland
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Adam Hartman, MD, FAAP, FANA, FAES - National Institute of Neurological Disorders and Stroke (NINDS), National Institutes of Health (NIH), Bethesda, Maryland	Steven Zullo, PhD - National Institute of Biomedical Imaging and Bioengineering (NIBIB), National Institutes of Health (NIH), Bethesda, Maryland