



## **Mitochondrial Medicine 2013: Newport Beach, CA**

June 12 – 15, 2013

### **The United Mitochondrial Disease Foundation (UMDF)**

#### **Final Report**

The United Mitochondrial Disease Foundation (UMDF) is proud to report on the success of the 2013 International Symposium held in Newport Beach, CA, June 12-15, 2013. UMDF's Symposium is the largest medical gathering devoted entirely to issues related to mitochondrial disease and welcomed medical professionals, patients, and their families from across the nation and around the world. The Symposium involved a four-day scientific session as well as a concurrent two-day family event. The scientific session provided CME credits through the Akron Children's Hospital.

#### **Sponsor Impact**

Symposium sponsors were prominently recognized with signage and table displays and in the banquet program with 400 guests in attendance. This year's registration included 240 researchers, physicians, and other allied health professionals and 154 individuals (including affected individuals and their families). Enclosed is an electronic copy of the Symposium syllabus, containing program topics, speaker information and a list of exhibitors and sponsors. Please also enjoy the enclosed thank you notes from our members whom wouldn't have been able to attend without the generous grants!

Significant time was provided during the final two days for doctors and scientists to interact with the family members who attended the Symposium. This is a special feature of the Symposium that brings together those affected with those treating them and seeking a cure.

#### **Unique and Valuable Programs**

UMDF continued to build upon the symposium's history of specialized programming this year. These programs were tailored for both affected families and the scientific community.

The family programs contained valuable sessions such as, "Diagnosis of Mitochondrial Disease – Past, Present and Future" with Richard Haas, MB, BChir, MRCP, "Understanding Seizures in the Mitochondrial Patient" with Russell Saneto, D.O., PhD., "Dysautonomia and Autonomic Issues in Mitochondrial Disease" with Richard Boles, M.D.

The scientific program was under the direction of Gregory M. Enns, MB, ChB, Associate Professor of Pediatrics, and has been Director of the Biochemical Genetics Program at Stanford University since 1998. He was trained in clinical genetics and clinical biochemical genetics at the University of California, San Francisco, graduating from the program in 1998. Wednesday's platforms included topics dealing with 'Mitochondrial Disorders in the 21<sup>st</sup> Century – Is Treatment Possible' and 'The Science Behind Clinical Management'. Thursday's program included topics of 'Mitochondrial Imaging' and 'Next Gen Reporters of Cellular Metabolism'. Friday's included platform topics of 'Animal Models and Translational Research' and 'Translational Mitochondrial Medicine'. Saturday's platform was dedicated to 'Bringing New Therapies to Market'.

Attendees of the programs, including patients, family members and members of the medical community thought very highly of the programming. A post-Symposium survey showed that the programming was well received and participants gave high praises to the symposium and UMDF saying, "The Doctor is in was wonderful and left me with so much hope because a doctor had more information for me than I've gotten in 3 years. I left feeling hopeful and motivated." "Attended my 9th symposium, always learn something new and meet up with new peers". Survey respondents were asked to rate the Symposium 1-5 (1=Poor 5=Excellent). The 2013 Symposium was rated very highly by attendants as either 5 (Excellent = 64.7%) or 4 (26.4%). The remaining responses rated the Symposium 3 (8.9%). No respondents rated the Symposium at the lowest two levels, 2 or 1.

## **Travel Scholarships**

A total of 83 individuals from 21 different states and 10 different countries were awarded travel scholarships totaling approximately \$43,980. Without this assistance, these UMDF members would not have been able to attend the symposium in Newport Beach, CA. In addition, UMDF assisted with travel costs for 31 speakers to attend the Symposium. All travel awardees were required to fill out an application for a scholarship. Based on the criteria needed for a grant, location, gender and economic structure, a team at the foundation read over to approve travel grants. Speakers who received funding included mitochondrial specialists such as, Dr. Bruce H. Cohen, Dr. Salvatore DiMauro, Dr. Marni J. Falk and many others. These speakers are chosen by the Scientific Planning Committee each year for their phenomenal work with mitochondrial diseases.

## **Research Awards**

Five researchers, whose projects may lead to a cure or better treatments for the thousands of adults, teens, and children who suffer from a mitochondrial disease, were awarded \$424,000 in research funds during the Symposium. This year's awards bring the total amount of UMDF-funded research to over \$10 million since 1996. The UMDF continues to be recognized as the largest, non-governmental contributor of grants focused on mitochondrial disease research. The money for research awards comes from special events and fundraisers held by Chapters and groups across the country. Following is a summary of the successful research proposals awarded funding at the 2013 Symposium (see page 4-5 of the Friday Night Banquet agenda):

**James Stewart, PhD.**, Max Planck Institute for the Biology of Ageing, Cologne, Germany. Chairman's Award Recipient - \$90,000 over 2 yrs.

"Using mtDNA mutator mouse-derived lineages to generate mouse models of human mitochondrial diseases."

By working with mice that are prone to mitochondrial mutations, Dr. Stewart will develop new genetic models of human disease. Once established, these mouse models can be used for the development of new drug therapies.

**Alberto Sanz-Monterro, PhD.**, University of Tampere, Finland.

\$100,000 over 2 yrs.

"A Genome-wide RNAi Screening to Identify New Genes Involved in Mitochondrial Disease."

Dr. Sanz-Monterro will use a well-understood fruit-fly model to discover previously unknown genetic defects that can cause mitochondrial disease. Many mitochondrial disease patients have not had a specific genetic mutation linked with their disease, and this research will help to fill that gap.

**Rajesh Ambasudhan, PhD.**, Sanford-Burnham Medical Research Institute, La Jolla, CA.

\$84,000 over 2 yrs.

"A Human Reprogrammed-Cell models of MELAS."

Dr. Ambasudhan will obtain skin cells from MELAS patients and reprogram them as nerve cells to be grown in culture. This "disease-in-a-dish" model will be used to gain insights into mitochondrial dysfunction in MELAS and other mitochondrial diseases.

**Natalie Niemi, PhD.** University of Wisconsin, Madison, WI.

\$75,000 over 2 yrs.

"Utilizing dynamically regulated phosphorylation as a means to modulate mitochondrial metabolism."

Dr. Niemi will study mechanisms that activate enzymes in the mitochondria, with the goal of understanding how this regulation is impaired in mitochondrial disease. This could lead to new therapeutic options for mitochondrial disease patients.

**Alicia Pickrell, PhD.**, National Institute of Neurological Disorders & Stroke, Bethesda, MD

\$75,000 over 2 yrs.

“Therapy for mitochondrial diseases: an investigation into the potential to stimulate Parkin-mediated mitophagy.”

Dr. Pickrell is studying the effects of the drug Rapamycin on the removal of abnormal mitochondria from cells in mice. This FDA-approved drug has the potential to selectively eliminate dysfunctional mitochondria in humans, helping to restore normal energy metabolism in mitochondrial disease patients.

### **UMDF Clinical Fellowship**

Along with the above five research grant awards, the UMDf also awarded its third UMDf Clinical Fellowship to **Amel Karaa, MD**, Harvard Medical School and Massachusetts General Hospital

\$70,000 over 1 yr.

“Hypogonadotropic hypogonadism in mitochondrial disease; prevalence, phenotypic heterogeneity and hormonal spectrum variations in a tertiary hospital cohort.”

The clinical fellowship training award is designed to support the training of physician scientists who plan to practice clinical management of patients with mitochondrial disorders and to conduct patient-oriented research in the field of Mitochondrial Medicine.

### **UMDF Awards**

Five of the organization’s national awards were presented to individuals at the Symposium’s annual banquet. Please see page 8-13 of the Friday Night Banquet agenda for further details and recognition of those individuals who have gone above and beyond to help UMDf and those affected by mitochondrial diseases.

### **Moving Forward**

UMDF is already planning for and looking forward to Mitochondrial Medicine 2014 in Pittsburgh, PA at the Sheraton Station Square. UMDf continues to thank all sponsors and individual supporters for their generous support this year and look forward to the great accomplishments these partnerships will be able to achieve in the future.