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Brighter Days: Mitochondrial Medicine in 2011

An Interview with Dr. Fran Kendall



[Sean Durity](#), Yahoo! Contributor Network

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Recently I sat down with Dr. Frances Kendall, a nationally beloved geneticist and mitochondrial specialist. Through her medical practice, [Virtual Medical Practice](#), she focuses her efforts on the clinical treatment of children and adults with mitochondrial diseases or other genetic disorders. These are the hard cases and the difficult diagnoses. She is also our daughter's mitochondrial specialist. She shared with me her views on these brighter days in mitochondrial medicine.

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Days of Knowledge

"There has been an exponential growth in the knowledge and understanding of mitochondria and [mitochondrial diseases](#) over the last two to three years," says Dr. Kendall. It is the most momentum she has seen in over twenty years in the field. That growth in knowledge was exhibited at the [2011 Mitochondrial Medicine Symposium](#) in Schaumburg, IL. This was the 13th annual gathering of researchers, doctors, patients, and families. Dr. Kendall, a panelist and moderator during the symposium, felt the high energy of the participants. That is exciting when talking about energy disorders!

The format of the symposium lends to the collaborative atmosphere. Rarely do physicians, researchers, patients, and families for any disease gather at one place. Dr. Kendall says that the interaction is crucial for mitochondrial medicine. The researchers doing the basic science work can "put reality into their research and theory." They realize the importance and urgency of their work. The physicians break out of the busyness of every day care to share new ideas and approaches with each other. For families, it is a unique opportunity to hear the latest news in mitochondrial medicine from the best minds in the field. And it is a time to build friendships with those who suffer similarly.

Days of Treatment

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Published by [Sean Durity](#)

Writing for real life impact, Sean is first a husband, then dad, then an Informix database administrator, Bible teacher, and

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Since there are [no cures for mitochondrial diseases](#), I asked Dr. Kendall what patients would find most hopeful from the conference. She quickly cited the successful Edison trial and the "forward

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momentum" in the field. Edison Pharmaceutical [presented results](#) on its trial compound, called EPI-743. The results included significant improvements in patients with specific mitochondrial disorders like Leigh's Disease and MELAS. Even more exciting is that several LHON ([Leber Hereditary Optic Neuropathy](#)) patients recovered their vision! The EPI-743 trial is a specific, though still limited, success in treatment. Dr. Kendall believes that the forward momentum in the field will help expand that single success into many more treatments in the years to come.

Days of Diagnosis

More successful treatments, though, will require specific diagnoses. In the recent past, patients were diagnosed after a muscle biopsy by describing which of the five biochemical processes (called complexes one through five) in the electron transport chain were not functioning properly. For example, our daughter was diagnosed with deficiencies in all five complexes. Dr. Kendall says that classification is no longer helpful in most cases. While two patients might have a Complex I defect, the genetic cause could be very different. Since the genes control the expression of different proteins, effective treatments would also be different.

There are new, non-invasive diagnostic tools. Three different labs (Medomics, Baylor, and Transgenomic) can use a blood sample to produce a DNA panel and screen hundreds of the 1500+ genes encoded for mitochondrial energy production. Dr. Kendall is encouraging all mito patients to have DNA panels completed. With these panels mito patients can be subdivided into groups based on the specific genetic cause for the disease. With a genetic diagnosis, patients are eligible for any treatment trials that arise for that specific diagnosis. Without a genetic diagnosis, patients are not going to be eligible for trials. To begin the process, talk with your doctor. The lab we chose is projecting that results will not be available for at least two to three months.

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Although there are so many exciting developments, a mitochondrial disease diagnosis is still devastating. There are no cures, and treatments may be years away. While Dr. Kendall expects

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"significant developments in treatments over the next five to ten years," that is a long time for families to wait. Some patients will not live that long. Still she encourages patients to embrace the longer term view. She agrees that this longer term outlook is difficult for struggling families to accept and celebrate. We would all love quick and easy answers, but mitochondrial medicine is a complex and ever-broadening field. There will not be a single treatment or cure for all.

The longer term presents another challenge. Dr. Kendall says that the greatest need in the field is for more mito-knowledgable physicians to care for patients. She has seen tragic situations where patients have suffered with no specific diagnosis (or even an incorrect one) for years. And the problem could get worse. Most of the leaders in the field are in their 50s, heading toward eventual retirement. She doesn't see a new generation of doctors taking up the challenge of these difficult diseases.

Days of Hope

The [United Mitochondrial Disease Foundation](#) is a primary sponsor for the symposium. It also funds basic and clinical research that will hopefully lead to effective treatments. This year, recognizing the same physician shortfall, it also funded the first recipient of its Clinical Fellowship Program, Dr. Anna-Kaisa Niemi, to see patients and conduct patient-centered research. The UMDF is helping push mitochondrial medicine further every year. It seems that the hard work and passionate fund-raising is beginning to bear fruit. Through all the difficulties of this disease, these are truly brighter days of hope.

More from this author:

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