


# Scientific Update

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One year ago, the National Institutes of Health, ML4 Foundation and Polycystic Kidney Disease Foundation held a joint three-day conference entitled Mucolin, Transmembrane Receptor Proteins and Human Disease. The conference was well attended by famous researchers from around the world. They represented the NIH, Harvard Medical School, Yale University, Johns Hopkins University School of Medicine, University of Arizona, Ohio State University, Texas Southwestern Medical Center, University of Maryland, University of Hawaii, Hadassah Medical School, University Freiburg (Germany), Universitat des Saarlandes (Germany), etc.

We learned that although the ML4 gene had been recently discovered, the function of its product, mucolin, was still unknown. A worm model of ML4 was described by Hanna Fares from the University of Arizona. The need for work on ML4 models in mice and fruit flies (*Drosophila*) was outlined. The conference generated much research interest among the attendees. A number of research teams have applied to the ML4 Foundation for seed money grants (\$30,000-\$50,000 per year for 2 years) in order to obtain preliminary data on ML4. This allows these researchers to subsequently apply to the NIH for more extensive financial support. Our Foundation has already been successful with this approach.

A number of articles on ML4 have recently been published, including:

1. Altarescu G, Sun M, Moore DF, et al. The neurogenetics of mucopolidosis type IV. *Neurology* 2002;59:306-313.
2. Bach G, Mucopolidosis type IV. *Molecular Genetics Metabolism* 2001;73:197-203.
3. Smith JA, Chan C, Goldin E, Schiffmann R. Noninvasive diagnosis and ophthalmic features of mucopolidosis type IV. *Ophthalmology* 2002;109:588-594.
4. Bargal R, Avidan N, Olender T, et al. Mucopolidosis type IV: novel MCOLN1 mutations in Jewish and non-Jewish patients and the frequency of the disease in the Ashkenazi Jewish population. *Human Mutations* 2001;17:397-402. 

## The Roscoe O. Brady Symposium

On October 8, 2002 the ML4 Foundation was invited to speak at the Roscoe O. Brady Symposium, in honor of the lifetime commitment Dr. Brady has made to the study of metabolic disorders. NORD (The National Organization of Rare Diseases) and the ML4 Foundation were the two groups represented at the symposium. This was a tremendous honor for the Foundation.

Randy Yudenfriend Glaser, the President of the Foundation attended the symposium and

addressed Dr. Brady and the conference. Below is a brief description of the words spoken:

*NIH Record, Dec 10, 2002, "Although many commented on Brady's research leadership and his impact on their work, several others--including Randy Yudenfriend, President of the Mucopolidosis IV Foundation, and Abbey Meyers, president of the National Organization*

*for Rare Disorders (NORD)--focused on his clinical side, describing how he has touched and changed the lives of his patients."*

*The Foundation would like to thank the NINDS/NIH for extending this invitation to us and giving us the opportunity to publicly thank Dr. Brady for his tremendous efforts and successes*