

SCHOOL OF MEDICINE

Department of Internal Medicine
Division of Rheumatology

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Mr. & Mrs. Steven Bevers
Energy 4 A Cure Foundation
5000 Eldorado Parkway
Suite 150, #403
Frisco, TX 75033

Dear Sharon and Steve,

Congratulations on raising \$36,000 via the golf tournament and an additional \$10,000 by your two nieces at their clay Pigeon Shoot in St. Joseph, MO. Much labor goes into this type of event and your Energy 4 A Cure Foundation gets it done each year. Thank you!

I want to report on several new developments. Parul Kothari, M.D. will be defending her Ph.D. thesis on February 24, 2014. She has been writing it this past month, working hard day and night and through the weekends. It covers clinical, radiologic and pathologic data on 11 afflicted families, basic research studies on TREX1 expression in various tissues but especially in the human brain, and progress towards developing a mouse model. Your gifts have supported all three of these aspects of her thesis. The funds are critical for this work to continue.

In carrying out these investigations, she is to be congratulated as she produced a key antibody to the TREX1 protein. This reagent has allowed us to substantially expand our studies. It has allowed her (and other research groups to whom we have provided this antibody) to definitively identify TREX1 proteins for the first time in human cells and tissues. The TREX1 protein is located inside cells. In the brain, one particular cell type (microglia) expresses relatively more of the protein, especially in response to injury such as a stroke.

We have recently identified a third St. Louis family with the disease. Assuming the St. Louis data can be applied nationally, there should be approximately 300 to 400 families in the United States with this disease. Family size varies from two to several hundred members in our cohorts so the disease process is not quite as rare as one might think. Similar numbers are available for the Netherlands (about one family/million people).

These advances in our understanding of the disease phenotype and the increased knowledge relative to the mutant proteins are supported by the funds you have raised.

Sincerely,

John

