On June 8, 2016, during my genetics rotation, Dr. Amato told me about a Grand Rounds conference hosted by Genetics and that Dr. Mabry would talk about Mabry syndrome. That was the first time I heard about Mabry syndrome, and my quick review revealed that the first-ever cases of Mabry syndrome were identified at the University of Kentucky (UK) in 1968, almost four decades prior to coining the term Mabry Syndrome. I volunteered my services, and I was given the opportunity to embark upon a journey of a lifetime with Dr. Mabry — visiting three original cases identified in 1968 and helping to collect blood samples and conduct a neurological assessment.
Our small team consisted of Dr. Mabry, Dr. Thompson (a research scientist from the University of Toronto working on Mabry Disease), Mr. Jim and myself. It was a two day trip, and we visited two different medical care facilities in Eastern Tennessee.

I got a chance not only to talk about Mabry disease but to also talk about Dr. Mabry’s 50 years of service at UK. Dr. Mabry told me about the history of the Department of Neurology, how Dr. Clark envisioned and started the department and how Dr. Clark encouraged Dr. Mabry’s son to start a career in neurology. Dr. Mabry recalled about the legislative challenges he faced when he introduced newborn state screen programs. In 1968, he received a call from a family physician, from his hometown in TN, about siblings and a cousin who had developmental delay and worsening seizures, and he needed Dr. Mabry’s help to identify the cause. Dr. Mabry saw those patients in his “GEM” clinic and found striking facial features, musculoskeletal findings and “incidental” findings of raised alkaline phosphatase. He recalled, “I did not order Alk Phosphatase. It was ordered as part of a metabolic panel, and when the results showed high Alk Phos values, I was clueless and did not know what to do but thought maybe there is some association. So I published my findings in the Journal of Pediatric with the title, ‘Familial hyperphosphatasia with mental retardation, seizures, and neurologic deficits.’”

In 1980 other researchers and geneticists were studying abnormal bone metabolism. Dr. Mabry explained the special phenotypical features and hyperphosphotases in a literature research found case series. Later, in 1990, six more cases from Europe were reported and then finally in late 1990s, genetic studies showed an autosomal recessive disorder of mental retardation and raised alkaline phosphatase. A group of scientists at the University of Toronto decided to honor Dr. Mabry by naming this syndrome after him as his case series resulted in the identification of this rare and new genetic disease and opened new horizons of genetic research.

To date, 30 cases of Mabry disease are identified and registered worldwide. Geneticists and researchers still use the Dr. Mabry report as one of the diagnostic criteria. Although retired in 2011, Dr. Mabry is still an enthusiastic, energetic and very engaged person, and he continues to contribute to his research. He is loved by his patients and their families and has impressive memory of naming almost all of his PKU patients by their first names; he proudly shares success stories of his patients. He is a living legend among us and continues to inspire young physicians.