THE TALE OF Mabry Syndrome

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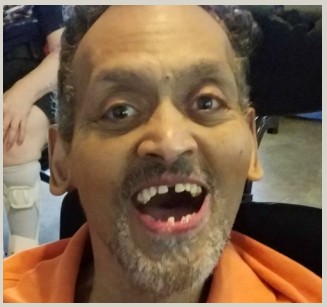
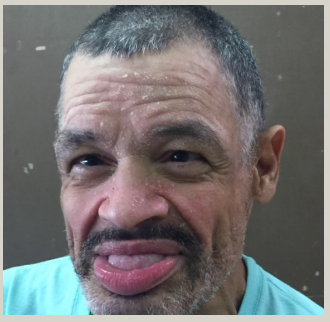


I am not in the picture.

I am not in the picture.

I am the one in the center.



I am standing with my mommy.

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 on 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 new born 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

UK'S FIRST NEUROLOGIST AND FOUNDER OF THE DEPARTMENT OF NEUROLOGY WAS CHILD NEUROLOGIST, VISIONARY AND FUTURIST DR. DAVID B. CLARK

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 hyperphosphotesia 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.

CASE HIGLIGHTS

CASE A: AGE 60

MEDICAL FACILITY RESIDENT

Seen by Dr. Mabry for the first time, his brother was originally seen by him in 1968. We had a detailed encounter and had an opportunity to talk to his caregivers helping us to know him better. He is **wheelchair-bound**, **non-ambulatory** and **G tube feeding dependent**. He is not taking any anti-seizure medicine and reportedly has had no seizure-like activity for the last eight years. He likes to listen to music and reportedly has been able to differentiate between different types of music and prefers contemporary pop over country music.

CASE B: AGE 58

MEDICAL FACILITY RESIDENT

Case B was seen by Dr. Mabry in 1968. He was 10 years old and was one of the first patients later given a clinical diagnosis of Mabry syndrome. He is cheerful and like his brother, he has a very **limited interaction** and has **compulsive behaviors** including chewing on hands and poking of the eyes, which resulted in elective left eye nucleation. As compared to his brother, he is ambulatory and has phenotypical features more suggestive of Mabry as found in other patients. He likes to go on picnics with his caregivers.

CASE C: AGE 64

MEDICAL FACILITY RESIDENT

Initially seen by Dr. Mabry in 1968 when he was around 17 years old, Case C is the oldest known patient with Mabry syndrome. He has limited interaction and does not like visitors. He is on Leviteracetam and reportedly has had no seizure-like activity for more than a year. He likes his toy horse and always keeps it with him. He **ambulates** and **tolerates pureed foods**. He also has phenotypical features as described in other cases of Mabry disease. He likes to go on picnics and likes animals.