Cardiologist transforms care of patients with rare heart disorder

October 9 2015, by Emily Boynton

In 1970, a woman with a history of fainting spells was referred to Arthur J. Moss, M.D., a young and well-regarded cardiologist at the University of Rochester Medical Center. Little did anyone know, including Moss, that this meeting was the start of a decades-long journey that would transform the diagnosis, treatment and quality of life of patients with a rare and potentially fatal heart rhythm disorder called long QT syndrome (LQTS).

In the September/October issue of *Progress in Cardiovascular Diseases*, Moss and his three adult children detail the events that led to manifold advances in LQTS, including: knowledge of risk factors that enable early diagnosis; the discovery of multiple treatment options that decrease the risk of sudden cardiac death; the creation of the International LQTS Registry, one of the first gene registries for any disease in the world; and the identification of sixteen genes (and counting) associated with the disorder.

"Dr. Moss is the world's foremost expert in long QT syndrome and one of the most influential scholars in the fields of electrophysiology and cardiology," said Carl "Chip" Lavie, M.D., Professor of Medicine, Ochsner Clinical School-University of Queensland School of Medicine in New Orleans, Louisiana and editor-in-chief of Progress in Cardiovascular Diseases. "I am honored to publish such a special paper showing the history of long QT syndrome and how the management of the disease has evolved through a coordinated effort that Dr. Moss has been intimately involved with over the past 45 years."

The disease, which affects approximately 50,000 people in the U.S., makes the heart particularly susceptible to arrhythmias – irregular heart rhythms that can trigger fainting spells and sudden cardiac death. When Moss, the Bradford C. Berk, M.D., Ph.D. Distinguished Professor in the Department of Medicine, evaluated his first patient in 1970 there were no drugs or devices available to prevent or stop the dangerous arrhythmic episodes.

Together with Joseph McDonald, M.D., then professor and chief of Neurosurgery at the University of Rochester Medical Center, Moss devised a surgical treatment (called a left-sided cervicothoracic sympathetic ganglionectomy) that turned out to be remarkably effective in limiting the dangerous arrhythmias his patient experienced. Moss and McDonald published the details of this therapy in 1971. A few years later, beta blockers – drugs that make the heart beat more slowly – came on the market. Moss and other physicians used these medications in patients with LQTS and found they were also beneficial.

In the years that followed, Moss received an increasing number of consultation requests involving patients with LQTS. The number of referred patients was more than he could manage, and in an effort to maintain contact, he established the Long QT Syndrome Registry in 1974.

Presently, there are more than 1,000 LQTS families from around the world enrolled in the registry and more than 2,500 affected family members. Moss' team contacts families once per year to discuss their overall health, medications, and fainting or arrhythmic episodes. This information is added to the registry and used by hundreds of scientists and physicians to further our understanding of the disorder.

The National Institutes of Health has supported the registry since its creation. In 2014, Moss received another NIH grant to fund the registry

and associated research projects through 2019. Though Moss' focus is on research, LQTS patients from all over the world continue to travel to Rochester for evaluation and treatment by Spencer Z. Rosero, M.D., Associate Professor of Medicine and director of the Hereditary Arrhythmias Clinic at UR Medicine's Strong Memorial Hospital.

"Helping patients with long QT syndrome has been a highlight of my career, and our work is not done yet," noted Moss. "It is really remarkable what can result from the effective treatment of a single patient. The research and practice of medicine is unpredictable, but always extremely rewarding."

Dr. Moss has followed his first patient, Helen Ruth Pontera of Canandaigua, New York, since the beneficial surgery was performed in 1970. Mrs. Pontera is in good health at age 85 without fainting spells. She thinks the world of Dr. Moss and considers him her doctor and her friend.

Moss' children – Katherine M. Lowengrub, M.D., Instructor in Psychiatry at the Sackler School of Medicine in Tel Aviv, Israel; Deborah R. Moss, M.D., M.P.H., Associate Professor of Pediatrics at the University of Pittsburgh Medical Center; and David A. Moss, Ph.D., Professor at Harvard Business School – interviewed their father over the past year to gather information for the commentary.

"My dad asks great questions and he doesn't stop until he figures a problem out. His perseverance, creativity and collaborative nature have yielded incredible results in LQTS," said his daughter Deborah. "But, most inspiring, is the unique relationship he developed with his patients: he understood the importance of really listening and building trust. Together, a doctor and a patient can be a powerful force for change."

More information: Katherine M. Lowengrub et al. "Long QT

Syndrome: How Effective Therapy in a Single Patient Favorably Influenced the Long-Term Clinical Course and Genetic Understanding of this Hereditary Disorder," *Progress in Cardiovascular Diseases* (2015). DOI: 10.1016/j.pcad.2015.08.002