For Immediate Release
Contact: Denise Portner
dportner@steegethomson.com | 267.670.7010

Mother/Daughter Team to Present Breaking Research at ANA2019 on Successful Targeted Treatment of Neuropathy in a Large Family

Appearance marks third generation of ANA presenters; A family helping families

Investigators Anne Louise Oaklander, MD, PhD; and daughter Madeleine C. Klein

May 2002: Isabelle Rapin, MD, and granddaughter Madeleine C. Klein, age 8

(MOUNT LAUREL, NJ, October 13, 2019) — When Madeleine C. Klein takes her place at today’s poster session of the 144th Annual Meeting of the American Neurological Association (ANA), she will be the third generation of women in her family to do so.

The pre-med student, together with her mother, Anne Louise Oaklander, MD, PhD, Associate Professor of Neurology at Harvard Medical School and Director of the Nerve Unit at Massachusetts General Hospital, are presenting competitive late-breaking abstract LBA-S313: “Normalizing Sodium-channel Firing in Small-fiber Neuropathy from SCN9A-variant May Improve Axonopathy Along with Symptoms.”

The paper describes the progress that Oaklander’s lab has made since 2013 in diagnosing and treating small-fiber polyneuropathy, which runs through the Soares family of Uxbridge, MA.
Through characterization of a mutation in the SCN9-A gene that controls a critical sodium channel in small nerve fibers, the team has been able to resolve symptoms in multiple family members. José and Sarah Soares first brought their son Sebastian to Dr. Oaklander in 2015 as a very sick 9-year old boy who cried constantly and couldn’t attend school. His symptoms included red, painful and itchy feet with ulcers, tachycardia, growth retardation, and secondary depression/anxiety.

Further sequencing revealed that Sarah, her father, one of Sebastian’s two brothers, and a 7-month-old sister also carry the same SCN9-A gene variant. The team treated symptomatic family members with currently available sodium channel blocking drugs, with startling results. Not only did the drugs almost entirely resolve the pain, but patients’ nerve endings regrew so successfully they were able to discontinue most medications.

“We went from hopeless to hopeful,” said Sara Soares, who had brought Sebastian to dozens of physicians before meeting Dr. Oaklander. “Our lives used to be so difficult, not knowing what would happen from one day to the next,” she said. Sebastian, now 14, attends school full time and “doesn’t have to wear sandals anymore,” Soares said.

His brother Oliver, 6, is also symptom free, she added.

“Small-fiber neuropathy is emerging as one of the most common neurological disorders, but many neurologists won’t see these patients, instead sending them to pain specialists without training in neurodiagnosis or awareness of therapies,” Oaklander said. “Accurate diagnosis and targeted treatment are more effective and safer than pain-killers.”

Klein, who earned a Bachelor of Science degree in Biology with honors from the University of Massachusetts at Amherst, says her primary interest is genetics. She developed her love of science at a young age, influenced by the careers of her mother and of her grandmother, the renowned neurologist and ANA Member Isabelle Rapin, MD.

Rapin, who passed away on May 24, 2017, at age 89, conducted research on childhood communication disorders at the Einstein College of Medicine in the Bronx, NY, for more than 50 years. A founder of the field of child neurology, she is credited with a number of discoveries in the fields of neurogenetic and communication disorders in childhood, including shaping our understanding of autism and helping popularize the concept of autism as a spectrum of disorders.

Klein shares her grandmother’s interest in pediatrics. She and her grandmother have both published papers on neuronal ceroid lipofuscinosis, Klein’s earlier focus. Earlier this month, she attended the inaugural Grand Rounds lecture series at Albert Einstein College of Medicine named for Rapin, whose career was there, and delivered by Oaklander. Klein’s father, Max Klein, PhD, an engineer, also works in the Oaklander lab; and her brother Daniel Klein, who holds a PhD in particle physics, works in Data Science for Amazon.
“Science is really a family business over here,” said Klein, who won a travel scholarship to present the poster at the ANA Meeting. “My family never shied away from talking about medicine at the dinner table. They’d talk about what they saw in clinic. It’s always been there for me.”

Through their current research on the SCN9-A gene variant, Klein said, “our family is working to help this family.”

Oaklander and Klein will present their late-breaking abstract **LBA-S313 on Sunday, October 13, from 5:30–7 pm. in the Majestic Ballroom at the Marriott St. Louis Grand (800 Washington Ave, St. Louis, MO).**

**About ANA2019**

The 144th Annual Meeting of the American Neurological Association (ANA) will be held October 13–15, 2019 at the Marriott St. Louis Grand, with the “Brain-Computer Interfaces in Neurological Disease” Pre-Meeting Symposium on October 12 on. More than 900 of the nation’s top academic neurologists and neuroscientists, as well as students, trainees, and international professionals, will convene to share three days of research at the forefront of neurology and neuroscience. Follow the meeting live using #ANA2019 on Twitter @TheNewANA1, on Facebook @AmericanNeurologicalAssociation, or on Instagram @ananeurology.

**About the American Neurological Association (ANA)**

From advances in stroke and dementia to movement disorders and epilepsy, the American Neurological Association has been at the vanguard of research since 1875 as the premier professional society of academic neurologists and neuroscientists devoted to understanding and treating diseases of the nervous system. Its monthly *Annals of Neurology* is among the world’s most prestigious medical journals, and the ANA’s *Annals of Clinical and Translational Neurology* is an online-only, open access journal providing rapid dissemination of high-quality, peer-reviewed research related to all areas of neurology. The acclaimed ANA Annual Meeting draws faculty and trainees from the top academic departments across the U.S. and abroad for groundbreaking research, networking, and career development. For more information, visit [www.myana.org](http://www.myana.org) or follow @TheNewANA1 on Twitter, @AmericanNeurologicalAssociation on Facebook, or @ananeurology on Instagram.