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ALUMNI PROFILE

Carolyn Macica, Ph.D. '96

Making Strides in Rare Disease, XLH

BY RHEA HIRSHMAN

On a Friday afternoon, teams of students at St. Martin de Porres Academy, a New Haven, Connecticut, middle school serving children from low-income families, gather for their science sessions. Supervised by medical and graduate school students from Quinnipiac University, the teams engage in what Carolyn Macica, Ph.D. '96, emphasizes is "real, hands-on scientific work" in disciplines ranging from chemistry to genetics. "We want to get kids excited about how the world around them works, and to introduce this population to the opportunities available in STEM fields," she says.

Dr. Macica is associate professor of medical science at Quinnipiac's Frank H. Netter School of Medicine and an adjunct assistant professor in the Department of Pharmacology at Yale School of Medicine. In addition to her involvement with Science Friday, which she founded in 2014, Dr. Macica volunteers every year as a judge for regional science fairs. Her enthusiasm for science is rooted in her childhood on her family's dairy farm in upstate New York where— when she was not doing chores—she was free to explore the natural world around her.

After graduating from the State University of New York (SUNY) at Potsdam with a major in chemistry and a minor in biology, Dr. Macica "fell in love with research" through working on transitional fetal circulation at Columbia University. Subsequently, she obtained both her master's and doctorate in pharmacology from New York Medical College (NYMC). With the support of her NYMC mentor, Wenhui Wang, M.D., professor of pharmacology, she went on to complete postdoctoral fellowships in pharmacology and endocrinology at Yale School of Medicine.

While she was investigating a peripheral nervous system disorder, her department chair suggested that she submit a grant to study X-linked hypophosphatemia (XLH)—a progressive, genetic condition in which the kidneys fail to process phosphate and vitamin D normally, resulting in excessive phosphate loss through the urine, known as phosphate wasting. Because the body needs phosphate for healthy bones, muscles and teeth, the condition presents in children as rickets, soft bones and skeletal deformities; and in adults as a progression of the childhood symptoms, plus manifestations including enthesopathy, which is the mineralization of the tendon insertions involving the upper and lower extremities and spine, resulting in painful, rigid bone spurs; early-onset osteoarthritis; fractures; dental problems and hearing loss.

Dr. Macica received the grant and gave up her peripheral nervous system work to focus on metabolic bone disorders, particularly XLH.

Now, Dr. Macica is recognized as the national expert on adult XLH, which she has published numerous papers, authored textbook chapters and given more than 30 research presentations on. While still focused on the basic science—using mouse models to examine how and why the disease's co-morbidities occur—she

has also undertaken clinical and translational research, applying laboratory findings to address the condition's implications for those who live with it.

"The impact of XLH on activities of daily living is profound," Dr. Macica says, "and few health care providers—including physical therapists—know how to work with these patients." In a study under her guidance, a team of Quinnipiac faculty from physical therapy, occupational therapy, diagnostic imaging and social work, conducted a comprehensive, interdisciplinary analysis of the disorder to evaluate the physical and functional impact of XLH in adulthood as well as quality of life concerns.

Then, using the clinical data and patient input, Dr. Macica and colleagues developed a model for a specialized translational physical therapy program, implemented remotely. "Because of the nature of XLH, we were not looking for significant gains in range of motion," she explains. "The goal was to help people increase engagement in their lives–whether walking the dog, cooking more or being able to maintain social connections."

The experiment worked, with participants showing marked improvement in all assessment areas and reporting increased confidence in their ability to navigate their lives. The team has submitted their findings to a peer-reviewed journal. Once published, the results will serve as an evidence-based physical therapy program that can improve quality of life for adults with XLH.

As chair of the scientific advisory board for the national XLH network, Dr. Macica, along with an interdisciplinary team, developed the first patient toolkit for managing XLH across the lifespan, addressing issues from communicating with health care providers to dating and family planning. While her primary focus remains phosphate-wasting disorders, Dr. Macica describes herself as "a passionate and vocal advocate for the rare disease community." She is the founder and chair of Netter's annual Rare Disease Day (RDD) Symposium—a CME-accredited event of scientific and patient-centered programming—and faculty advisor for the student chapter of the National Organization for Rare Diseases (NORD).

"Being able to directly apply research to evidence-based outcomes is not always possible for a scientist," Dr. Macica says. "I have been blessed to be able to move between bench and bedside." ■

MILESTONES
Alumni Achievements

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The 10s

Tina Fickeria, M.S. '18, was promoted to Research and Development Specialist at Regeneron Pharmaceuticals. She is the program lead for numerous immuno-oncology treatments and bispecific antibody therapies.

Zachary Ehrlich, M.P.H. '17, was promoted to epidemiologist with the New Jersey Department of Health in May 2021, and began a Doctor of Public Health (Dr.P.H.) program in leadership, practice and research at Rutgers University in the fall of 2021. He also worked with the National Environmental Health Association as a subject matter expert and contributor for *A Guide for Environmental Health Responsibilities and Competencies*, fifth edition, released in June 2021.

Michael V. Longo, M.D.'15, joined Rhode Island Medical Imaging, a network of 12 private, state-of-the-art medical diagnostic imaging facilities, as a radiologist. He is a member of the American Medical Association, Radiologic Society of North America, American College of Radiology, Society of Abdominal Radiology and the Society of Radiologists in Ultrasound.

Danielle Clark, M.D. '14, associate program director and assistant professor of medicine at the University of Cincinnati Medical Center (UCMC), was awarded the 2022-24 Jeremiah A. Barondess Fellowship in the Clinical Transaction from the New York Academy of Medicine, in collaboration with the Accreditation Council for Graduate Medical Education. The two-year, $50,000 fellowship will enable Dr. Clark to implement an educational initiative at UCMC aimed at improving patient-centered bedside rounds in the COVID-19 era.