## **UAMS News Bureau**

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## NIH Awards UAMS \$1.54 Million To Tackle Brittle Bone Disease

LITTLE ROCK — A UAMS research team is using a \$1.54 million grant from the National Institutes of Health (NIH) to support its goal of finding better treatments for osteogenesis imperfecta (OI), also known as brittle bone disease.

The four-year award from the NIH National Heart, Lung, and Blood Institute will allow Roy Morello, Ph.D., and his interdisciplinary team to test whether lung abnormalities in OI patients can be treated separately from OI's bone-related defects.

An estimated 25,000 to 50,000 people in the United States live with OI, a rare genetic disorder of connective tissues that has no cure. It is mainly caused by mutations in the genes responsible for producing Type I collagen, a protein that normally holds bones together and makes them strong. With inadequate collagen, bones become brittle and prone to fractures.

"Collagen is a primary building block for bones, and OI is like the perfect storm for the skeleton," said Morello, associate professor in the College of Medicine departments Physiology & Cell Biology, Orthopaedic Surgery, and the Division of Genetics.

OI also plagues other organs and tissues, including the lungs, skin, teeth and heart, and it affects hearing. In severe cases, the disease can be lethal in infants.

"Collagen really covers a broad spectrum of issues that these patients unfortunately are subjected to," Morello said. "There is obviously a desperate need for new therapies."

The impact of OI on respiration has long been thought to result from the OI-caused bone issues, such as chest wall deformities and abnormal spine curvature, but recent studies by Morello's team challenge that view. The findings have led to their hypothesis that respiratory defects in OI patients could be treated separately from their skeletal fragility.

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"We were among the first to show that there were primary changes in the lung caused by mutations in Type I collagen," Morello said.

He is excited to have developed a novel method for testing the genetic mutations on specific tissues such as the lungs.

"This is a completely new model to study the disease, and it should lead us to a much better understanding of OI," he said. "Our long-term goal is to identify new treatments for the respiratory disease and prevent disability and death from OI."

Collaborators on the research project include John L. Carroll, M.D., a professor and section chief for the Pediatric Pulmonary and Sleep Medicine Division in the College of Medicine Department of Pediatrics.

"We are fortunate to have Dr. Carroll, with his great pulmonary expertise, join our team," Morello said.

Other collaborators are:

- Intawat Nookaew, Ph.D., associate professor, College of Medicine Department of **Biomedical Informatics**
- Milena Dimori, Ph.D., DVM, research associate and lab manager
- Anne-Karina Perl, Ph.D., Cincinnati Children's Hospital Division of Pulmonary Biology
- Sergey Leikin, Ph.D., senior investigator, NIH Section on Physical Biochemistry •

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Morello's work leading to the NIH award has been supported by a previous NIH award, an American Lung Association Innovation Award, the Osteogenesis Imperfecta Foundation and the UAMS Center for Musculoskeletal Disease Research, which is funded by the NIH Center of Biomedical Research Excellence.

UAMS is the state's only health sciences university, with colleges of Medicine, Nursing, Pharmacy, Health Professions and Public Health; a graduate school; a hospital; a main campus in Little Rock; a Northwest Arkansas regional campus in Fayetteville; a statewide network of regional campuses; and seven institutes: the Winthrop P. Rockefeller Cancer Institute, Jackson T. Stephens Spine & Neurosciences Institute, Harvey & Bernice Jones Eye Institute, Psychiatric Research Institute, Donald W. Reynolds Institute on Aging, Translational Research Institute and Institute for Digital Health & Innovation. UAMS includes UAMS Health, a statewide health system that encompasses all of UAMS' clinical enterprise. UAMS is the only adult Level 1 trauma center in the state. UAMS has 3,275 students, 890 medical residents and fellows, and five dental residents. It is the state's largest public employer with more than 12,000 employees, including 1,200 physicians who provide care to patients at UAMS, its regional campuses, Arkansas Children's, the VA Medical Center and Baptist Health. Visit www.uams.edu or www.uamshealth.com. Find us on Facebook, X (formerly Twitter), YouTube or Instagram.

