

BASIC//DISCOVERY

A DISTINGUISHED CAREER HONORED—

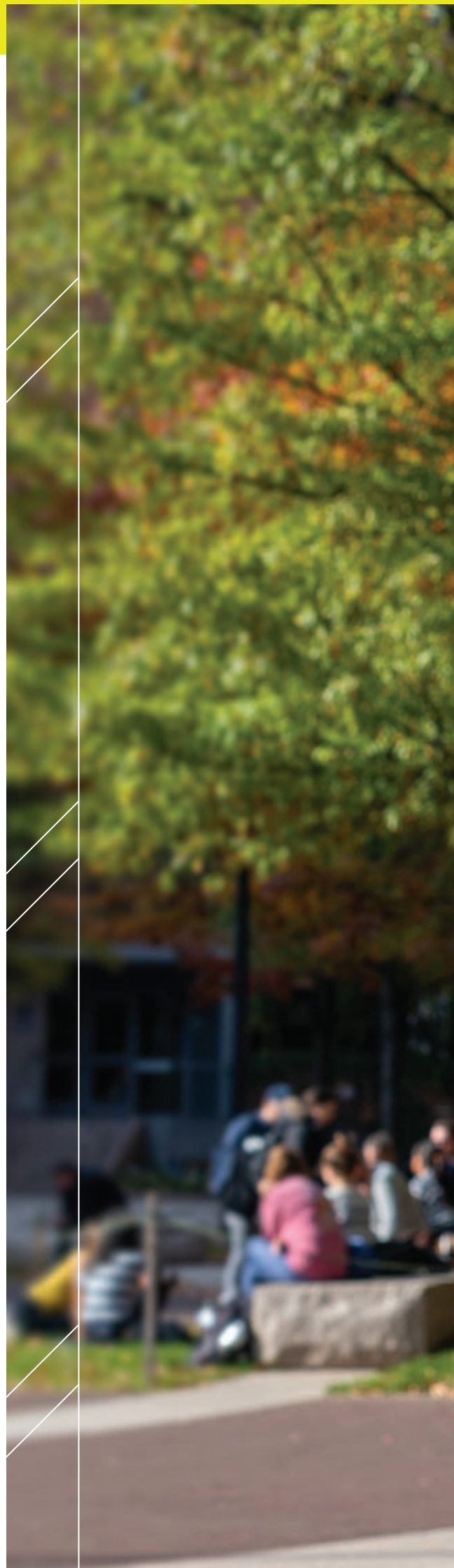
And the Work Continues

THE MOST PRESTIGIOUS AWARD IN INTERNATIONAL DERMATOLOGY—

the World Congress of Dermatology's Alfred Marchionini Medal in Gold—is bestowed every four years, and in 2019, **Jouni Uitto, MD**, professor and chair of dermatology and cutaneous biology, was the humble recipient. The award is a recognition of Dr. Uitto's long and distinguished career, in which he has published nearly 900 peer-reviewed articles, textbook chapters and reviews—including 30 new publications over the past year.

Dr. Uitto is particularly renowned for his research on a group of genetic diseases called epidermolysis bullosa (EB), which manifest at birth with extensive skin blistering and scarring. In its most severe form, EB can be fatal during a child's first weeks or months of life. His lab discovered the disease's genetic basis: mutations in the collagen7A1 gene undercut the production of a protein necessary for keeping layers of the skin together. The lab then developed a transgenic mouse model of EB, which became a platform for development of treatment approaches. Today, five gene therapy approaches in clinical trial have their origins in Dr. Uitto's work.

He is also driving advances on the rare childhood disease pseudoxanthoma elasticum (PXE)—where mineral build-up in elastic fibers of the skin and blood vessels leads to formation of plaques in blood vessels and eyes that cause heart attacks and blindness. Collaborating closely with parents of children with PXE, Dr. Uitto gathered DNA samples that enabled his lab to identify the responsible genes. Today, clinicians can make a prognosis based on a patient's specific set of mutations, and research continues to develop and test effective therapies. ■



Jouni Uitto, PhD
Professor and Chair of Dermatology
and Cutaneous Biology

