



COLUMBIA

CENTER FOR PRECISION
MEDICINE AND GENOMICS

IN THE DEPARTMENT OF MEDICINE

Internal Medicine Genetics Case Studies

“VEXAS Syndrome: Evolving Understanding of a Multi-Organ Inflammatory Disease”

VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome is a rare late-onset autoinflammatory disorder caused by somatic mutations in the UBA1 gene. Although most reported cases of VEXAS manifest as severe multi-organ inflammation, we will present a case with atypical clinical manifestations. We will then discuss recent mechanistic insight into this mysterious disorder, diagnostic approaches, and advances in therapeutic management.

Yiming Luo, MD, MHS

Assistant Professor of Medicine
Department of Medicine
Division of Rheumatology

Maria Salgado Guerrero, MD

Assistant Professor of Medicine
Department of Medicine
Division of Rheumatology

Friday, Dec. 6th, 10am

William Black Building
BB15-1514