

F. SESSIONS COLE, M.D.

Brief Bio, 2027 Mary Ellen Avery Award

F. Sessions Cole, M.D., is Professor of Pediatrics, Emeritus, at Washinton University in St. Louis School of Medicine. He is internationally recognized for his work on identifying genetic causes of pulmonary surfactant deficiency which disrupt fetal-neonatal pulmonary transition and for ending diagnostic odysseys in infants, children, and adults with other rare genetic diseases. Early in his career, he participated in the discovery of the first monogenic cause of progressive, lethal respiratory failure in term infants (surfactant protein B deficiency) and its treatment with infant lung transplantation based on a single family with 3 affected siblings. Since then, using NIH-supported state-of-the-art sequencing strategies, computational analysis platforms, cell-based and model-organism based functional characterization of rare genomic variants, and team science in the NIH-funded Undiagnosed Diseases Network, he expanded diagnosis of genetic causes of neonatal respiratory distress and described more than 60 genetic diseases in infants, children, and adults with diverse undiagnosed phenotypes. He has also led efforts to integrate affected individuals and their families as partners in NICU-based research, clinical care, and education, to develop strategies to prioritize mechanism-based, patient-informed therapeutic strategies for ultra-rare conditions, and to integrate exposome triggers into evaluations of undiagnosed individuals. His contributions have led to his appointments to serve on the National Advisory Child Health and Human Development Council for the Eunice Kennedy Shriver National Institute for Child Health and Human Development (subsequently rescinded by the current NIH Director), on the National Board of the March of Dimes, as Chair of the Board of Directors of the Undiagnosed Diseases Network Foundation, on multiple Institute of Medicine (now National Academy of Medicine) Committees, and on the FDA's Risk Communication Advisory Committee. His commitment to collaboration and mentorship has helped support the success of the UDN, successful integration of genomic medicine into NICU care, and success of his mentees as they have pursued leadership positions in academic Neonatology.