

Marshall L. Summar, MD,

Director: Rare Disease Institute
Chief, Genetics and Metabolism
Margaret O'Malley Chair of Genetic Medicine
Children's National Hospital, Washington, D.C.
Professor of Pediatrics, George Washington School of Medicine

Wikipedia: https://en.wikipedia.org/wiki/Marshall_Summar

Education:

Vanderbilt University, BS in Molecular Biology, 1981
University of Tennessee Center for Health Sciences, MD, 1985
Vanderbilt University Medical Center, Pediatrics Residency, 1988
Vanderbilt University Medical Center, Genetics Postdoctoral Fellowship, 1990

Boards:

American Board of Pediatrics
American Board of Medical Genetics both Clinical and Biochemical Genetics

Research and Development:

170+ peer-reviewed publications
25+ funded research projects (NIH, industry, and philanthropy)
100+ International/National invited talks
60+ Issued Patents

Current Leadership Roles:

National Organization for Rare Disorders, Board Chairman (2016-2020)
PHLOW Pharmaceuticals (Public Benefit Corporation), Board of Directors (2020-Present)
Arkansas Children's Hospital Research Institute, Board of Directors (2019-Present)
Black Women's Health Imperative, Rare Disease Diversity Coalition, Advisory Board and Research Co-Chair (2019-Present)
National Organization for Rare Disorders, Chair Scientific and Medical Advisory Committee (2015-Present)
Society for Inherited Metabolic Disorders, Past-President (2014-18)
Patient Centered Outcomes Research Institute, Chairman Rare Diseases Advisory Board (2013-2015)
National Urea Cycle Disorders Foundation, Scientific Advisory Board (1994-Present)
Chair, Clinical Advisory Board, Hemoshear Therapeutics (2016-Present)
Global Commission to Reduce the Time to Diagnosis in Rare Disease, Commissioner (2018-Present)

245 words

Dr. Summar is well-known for his pioneering work in caring for children diagnosed with rare diseases. He joined Children's National in 2010 from Vanderbilt University. At Children's National he leads the Division of Genetics and Metabolism, currently the largest known clinical division seeing over 8000 patients a year with rare diseases. Dr. Summar's laboratory works on both devices and treatments for patients with genetic diseases and adapting knowledge from rare diseases to mainstream medicine. His work has resulted in new drugs in FDA trials for patients with congenital heart disease. His laboratory is best known for its work in the rare diseases affecting nitrogen and ammonia metabolism. Dr. Summar has also organized and led a large number of international work groups to develop standards of care and treatment for rare diseases resulting in significant improvements in outcomes. He has been listed with Best Doctor's in America since 2004. He is the past Board Chairman for the National Organization for Rare Disorders and currently chairs their Scientific and

Medical Advisory Committee. He is very active in newborn screening issues developing testing and follow-up systems. He has developed and launched at Children's the world's first Rare Disease Institute. This RDI focuses on developing the clinical care field of the over 7000 rare diseases currently recognized. The RDI is the first Clinical Center of Excellence designated by NORD and focuses on building best clinical practices and diagnostic pathways.

88 Words

Dr. Summar joined Children's National in 2010 where he built and leads the Rare Disease Institute. He is best known for his work in developing registries and treatment standards for rare diseases. His laboratory work currently focuses on filling gaps in knowledge and testing for biochemical rare disorders. He is the past National Organization for Rare Disorders Board Chairman and helps develop NORD programs around natural history studies, undiagnosed disorders, and clinical centers of excellence in rare disease.

Photographs



