

Finding the Cause of Congenital Diarrhea



James Goldenring

Paul Sanger Professor of Surgery, Professor of Cell and Developmental Biology, Co-Director, Epithelial Biology Center

CURRENT RESEARCH

Early diagnosis and treatment of infants with deadly congenital diarrheal disease

Each year children are born with deadly diarrheal syndromes with no discrete cause. It is estimated that 5–10 of these infants are cared for each year by every newborn intensive care unit in the United States and likely throughout the world. Because no discrete diagnosis can be discerned, these children often spend months in newborn intensive care units sustained by intravenous nutrition. Ultimately they often die from sepsis or failure to thrive. Dr. James Goldenring, of Vanderbilt University School of Medicine, is seeking to understand the molecular bases of congenital diarrheal diseases that could be targets for potential treatments and thus, decrease the profound morbidity and mortality observed in these children. Dr. Goldenring's goal in the case of neonatal diarrheas is to change the standard of care and provide more rapid identification of the genetic causes of congenital diarrheas that can lead to early and effective interventions to ameliorate these often deadly diarrheal syndromes.

Over the past year, Dr. Goldenring led a group of physicians and scientists to impact the care of infants with congenital diarrheal disease without diagnosis. By employing state-of-the-art genomic sequencing analysis of the affected children and their parents, his group has successfully identified the genetic mutations responsible for diarrhea in the first four patients studied. These have led to alterations in diet that have cured one patient, strongly improved the health of two others, and eliminated the need for and risk of intravenous feeding. Unfortunately, insurance companies presently do not pay for whole genome analysis, and therefore there is a great need to establish a new standard of care. As these...

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AFFILIATION

Vanderbilt University

EDUCATION

- M.D. in 1986
Yale University School of Medicine
- Ph.D. in Molecular Biophysics and Biochemistry 1984
Yale University
- A.B. magna cum laude in Biochemical Sciences 1980, Harvard University

AWARDS

- American Association for the Advancement of Science Fellow, 2014
- Association of American Physicians, 2013
- Takeda Distinguished Research Award
- American Gastroenterological Association Fellow, 2006
- American Society for Clinical Investigation, 2004

RESEARCH AREAS

Life Science, Diagnostics, Genomics / Congenital, Oncology / Cancer

FUNDING REQUEST

Your contributions will support the continued research by Dr. James Goldenring at Vanderbilt University School of Medicine, as he seeks to understand the molecular bases of congenital diarrheal diseases that could be targets for potential treatments. Funding will support the \$2M required to accomplish more rapid genetic analysis and to understand and treat the genetic causes of diarrhea. In choosing to support his research, you will play a role in improving the health of the many children affected by congenital diarrheal diseases.