

## PEDIATRIC GRAND ROUNDS

Wednesdays at 8 a.m. in the Children's auditorium. For CME information, contact the UT Southwestern continuing medical education at 214-648-2166.

Sept 6

**"Fetal Origins of Hypertension and Renal Disease,"** Michel Baum, M.D., Professor and Director, Division of Pediatric Nephrology, UT Southwestern.

Sept 13

**"Pediatric End-of-Life Care: Torpedoes or Hugs?"** Robert Bash, M.D., Assistant Professor of Pediatrics, Division of Hematology-Oncology, UT Southwestern.

Sept 20

**"The Wiskott-Aldrich Syndrome - More Than a Rare Disease,"** Hans Ochs, M.D., Professor of Pediatrics, Division of Infectious Diseases, Immunology, and Rheumatology, University of Washington School of Medicine, Seattle.

*Also available via videoconference at selected area hospitals. Call 214-345-2330 for locations.*

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## MEDICAL UPDATE: The primary immunodeficiency diseases

The primary immunodeficiency diseases are a group of heterogeneous disorders in which over 100 different genes have now been recognized. The prevalence is close to 1:10,000; however, because of incomplete reporting to existing registries, the true prevalence of symptomatic immunodeficiency is probably higher, at least 1:5000. The overall 1:10,000 incidence is one-fourth that of cystic fibrosis (1:2500), one-half that of congenital hypothyroidism (1:5000) and somewhat more common than phenylketonuria (1:14,000). According to these figures, there are approximately 400 new cases of primary immunodeficiency in infants born in the US each year (4 million live births).

These diseases are recognized in association with recurrent or persistent infection, infections with unusual pathogens or unusual presentation of common diseases. Furthermore, patients with primary immunodeficiency appear to have an increased predisposition for the subsequent development of cancers and autoimmune diseases. Because the immunodeficiencies are congenital and hereditary, most newly diagnosed patients are infants and children. However, as we live in an antibiotic era, many of these children may survive into young adolescence and adulthood before a diagnosis is made.

Clinical presentations of immunodeficiency include recurrent ear and sinus infections, pneumonias, failure to gain

weight and thrive, deep skin or organ infections, persistent or "difficult to treat" thrush, need for intravenous antibiotics to clear infections and a family history of immunodeficiency.

While there may not be a particular phenotype, physical signs of immunodeficiency include failure to gain weight, absent tonsils, persistent thrush, difficulties with healing, rashes and eczema that do not resolve with conventional therapies, severe warts, gum disease, cutaneous granulomas, an enlarged liver or spleen, telangiectasias in eyes and ears, ataxia, dwarfism, cartilage abnormalities and oculocutaneous albinism or generalized lymphadenopathy with hepatosplenomegaly may indicate an immunodeficiency. Particular syndromes also are associated with immunodeficiency diseases such as DiGeorge syndrome, velocardiofacial syndrome, Bloom's syndrome and ataxia telangiectasia, among many others.

Early and prompt diagnosis is essential and potentially life-saving. Therapies such as bone marrow transplantation and gammaglobulin treatments are available, and if a molecular diagnosis is established, prenatal diagnosis can be offered to parents and family members.

Contact Dr. M. Teresa de la Morena at [maite.delamorena@utsouthwestern.edu](mailto:maite.delamorena@utsouthwestern.edu), for more information on immune-related disorders.

## FACULTY UPDATE: Doug Baker named Director of Medical Services

Doug Baker, M.D., has been named Director of Medical Services and Interim Chief of Emergency Medicine at Children's. Dr. Baker will provide leadership and direction to the clinical medical services and will assist in the oversight and development of pediatric emergency services.

Prior to joining Children's and UT Southwestern, Dr. Baker served as Chief of Pediatric Emergency Services at Yale-New Haven Children's Hospital and Chief, Section of Pediatric Emergency Medicine, Department of Pediatrics at Yale University, School of Medicine.

Through his career, Dr. Baker has been a leader in the field of pediatric emergency medicine as a distinguished clinician, educator and clinical scholar. He has received a number of awards including the notable Jean A. Cortner Divisional Teaching Award for Excellence at Children's Hospital of Philadelphia for six consecutive years and the National Institute of Emergency Care Instructional Excellence Award.

Dr. Baker holds a doctorate of medicine from Temple University, School of Medicine and a Bachelor of Arts from Franklin & Marshall College. He was a chief resident at Children's National Medical Center, a fellow in Emergency Medicine at Boston Children's Hospital, and has held faculty positions at Johns Hopkins, the University of Pennsylvania and Yale. He is board-certified in both pediatrics and pediatric emergency medicine.