

GRAND ROUNDS

Sept 12

Ethics Lecture: Pediatric Chronic Pain: A Mind-Body Perspective

Lonnie Zeltzer, MD, Professor of Pediatrics, Anesthesiology, Psychiatry and Biobehavioral Sciences, David Geffen School of Medicine at UCLA, Director, Pediatric Pain Program, UCLA Mattel Children's Hospital, Associate Director, Patients and Survivors Program, UCLA Jonsson Cancer Center, Los Angeles, CA.

Sept 19

Clinical Intelligence: Population Health Monitoring

Donna Futterman, MD, Professor of Clinical Pediatrics, Director Adolescent AIDS Program, Children's Hospital at Montefiore Medical Center, New York, NY.

Sept 26

Development of the Pediatrician: The Complexity of Education in the 21st Century

George Lister, MD, Professor and Chairman, Department of Pediatrics, UT Southwestern Medical Center at Dallas, Pediatrician-in-Chief, Children's Medical Center at Dallas.

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MEDICAL UPDATE: Severe Combined Immunodeficiency (SCID)

Severe combined immunodeficiency (SCID) is a fatal disease if not treated early. At least 12 genes have now been recognized as causes of SCID. While the overall frequency is estimated to be 1 in 50,000 to 1 in 100,000 live births, the exact numbers remain unknown as no newborn screening program is available except for a recent pilot study pending initiation in Wisconsin. Recognized by the media as the "Boy in the Bubble" disease, children with SCID have severe T⁻ lymphocyte defects and succumb to common infections if left untreated. Early diagnosis is essential as optimal definitive therapies are available.

The clinical presentation of children with SCID is characterized by early onset of recurrent or severe infections. Chronic diarrhea, interstitial pneumonitis, persistent candidiasis, and failure to thrive are classic presentations. Opportunistic infections such as *Pneumocystis Carinii* and *Aspergillus* are not rare. Disseminated disease after BCG vaccination is fatal, progressive vaccinia after small pox and vaccine associated polio encephalitis were common vaccine associated complications. Viruses such as adenovirus, cytomegalovirus, parainfluenza virus, rotavirus and respiratory syncytial virus are common causes of morbidity and mortality.

Physical signs can include poor growth, absent tonsillar tissue, and in some children a diffuse erythematous rash secondary to graft versus host disease (GVHD). This form of GVHD is due to maternal T cell engraftment. These alloreactive maternal T cells cross the placenta during delivery and as they expand are responsible for the skin and systemic manifestations of GVHD, including hepatitis with hepatomegaly. The transfusion of unirradiated blood products to a child with SCID may cause an overwhelming proliferation of alloreactive T lymphocytes present in the blood product and cause rapidly fatal GVHD.

The most important laboratory finding in children with SCID is lymphopenia. The absolute lymphocyte count in a healthy child less than 1 year of age is between 3000 to almost 9000 cells/microliter. In fact, children with SCID could be recognized in the newborn period by the general pediatrician by obtaining either a cord blood or peripheral blood CBC with a differential. Eosinophilia and elevation of transaminases are common as well.

Diagnosis is made by measuring immunoglobulin levels and analyzing the different subpopulations of lymphocytes. Based on these studies, patients can be classified into those with absence of both T and B lymphocytes (T⁻/B⁻ SCID) or those without T cells but presence of B cells (T⁻/B⁺ SCID). In the latter group these B cells are non functional. Molecular analysis should be attempted in every case of SCID. In fewer than 10% of cases the molecular diagnosis is unknown. The most common form of SCID is the X-linked T⁻/B⁺ form.

The first and successful bone marrow transplant in the world was performed in a child with SCID and remains the only cure for these patients. Children with SCID can be transplanted with stem cells from either: a parent, sibling, cord blood or a matched unrelated individual from the general population. These stem cells will give rise to functional T and B lymphocytes that allow an individual to fight infections. More importantly, the earlier a transplant is performed, the higher the chances for survival and cure.

To learn more about immune related disorders please contact:
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