Mattel Children's Hospital UCLA

Immunodeficiency diseases in children more common than previously thought

In recent years, advances in immunology have contributed to an enhanced understanding of primary immunodeficiency disease (PID) and a greatly expanded number of disease-related genes. Dozens of major and minor immunodeficiency illnesses are now identifiable in children.

UCLA's Division of Pediatric Allergy & Immunology is a leader in the management of primary immunodeficiency disorders and has established diagnostic and treatment paradigms to improve patients' quality of life.

Evaluation for chronic infections

Children are often referred for evaluation of chronic respiratory infections, including sinusitis, otitis media and pharyngitis, and ongoing reliance on antibiotics. A careful diagnostic process is necessary to diagnose primary immunodeficiency disease.

More than half of children referred to the clinic do not have a primary immunodeficiency disorder. Instead, chronic infection is the result of the child's environment, allergies or lack of response to specific antibiotics. In rare cases, a patient is diagnosed with a systemic disease such as cystic fibrosis or gastroenterologic disease.

Personalized diagnostic services and treatment

"UCLA's weekly immunodeficiency disorders clinic was established to meet the specific needs of pediatric patients," says E. Richard Stiehm, MD, professor emeritus of pediatric allergy & immunology and a national authority on primary immunodeficiency disorders. Because many primary immunodeficiency disorders are hereditary, UCLA also provides genetic testing and counseling.

"We see a lot of these patients, so we recognize these disorders," he says. "We have specialized laboratory tests that can identify these disorders. We have a clinic to give intravenous immunoglobulin therapy, which is not an easy therapy to administer. We also work closely with the division of hematology/oncology to perform stem cell transplants."

Children who are not diagnosed with an immunodeficiency disorder can benefit from an evaluation to determine the cause of frequent infections and reduce over-reliance on antibiotics. "It's crucial to identify children who aren't getting appropriate treatment," says Dr. Stiehm.

A full compendium of diagnostic tests are available to evaluate the patient's immune system including:

- Complete blood count
- Immunoglobulin levels
- Flow cytometry for various lymphocyte subsets
- Antibody tests for response to vaccines
- Measurements of lung function (spirometry)
- Allergy skin testing
- Blood tests for IgE antibodies to allergies
- Imaging: chest and sinus X-rays and CT scans
- Food and drug challenges

Primary immunodeficiency disorders

More than 200 genetic variants of primary immunodeficiency disease can involve any component of the immune system, including the antibody, cellular immune, phagocyte and complement systems.

Antibody and leukocyte disorders are the most common types of disease:

- Selective antibody deficiency
- Selective IgA deficiency
- Hypogammaglobulinemia
- Neutrophil defects
- Transient hypogammaglobulinemia of infancy

Some of these conditions are hereditary, and diagnostic assessment includes a family history and necessary laboratory tests. Symptoms often include bacterial or fungal infections with unusual organisms or severe and recurrent infections with common organisms. For children with suspected immunodeficiency diseases, a key diagnostic procedure is to assess their antibody responses to vaccination prior to beginning therapy.

Treatment for immune deficiencies includes antibiotics and immune globulin infusions. The latter can be given either intravenously or subcutaneously but must be closely monitored for side effects. Immunoglobulin infusions are given under careful supervision, first in the clinic and later at home. Psychosocial support is available to help children cope with the demands of regular clinic visits.

Families are counseled on preventing infections; participation in daycare, school and athletics; use of mass transit and childhood vaccination.

DiGeorge syndrome and SCID

DiGeorge syndrome is the most common genetic disorder following Down syndrome. Babies often present with severe infections in conjunction with cardiovascular defects and endocrine abnormalities. At UCLA, children with DiGeorge syndrome are provided comprehensive treatment by a coordinated, multidisciplinary team.

Severe Combined Immunodeficiency (SCID) is a primary immunodeficiency disorder characterized by a severe defect in both the T- and B-lymphocyte systems that results in one or more serious infections within the first few months of life. UCLA is one of four referral centers in California for SCID-diagnosed infants identified by newborn screening. Early treatment with stem cell transplantation often results in a cure.

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