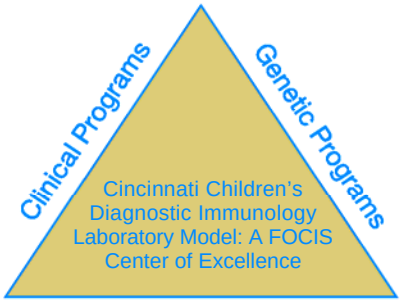
## Triangle of Immunology at Cincinnati Children's




Cincinnati Children's Diagnostic Center for Heritable Immune Deficiencies offers comprehensive immunologic and genetic testing, diagnosis, interpretation and treatment. Through an innovative program structure, physician-led teams from our laboratory, clinical care and human genetics areas collaborate to determine an accurate diagnosis, genetic implications and an optimal course of treatment for patients. This approach, which incorporates the expertise of physicians and researchers focused on specific areas within immunology, gives patients the best opportunity to improve outcomes.

# Enhance Pediatric Patient Care with Comprehensive Diagnostics for Immunological Diseases

During a recent interview, Alexandra (Lisa) Filipovich, MD, a leading expert in the field of diagnostic immunology who serves as the director of the Immune Deficiency and Histiocytosis Program, discussed the development of the Diagnostic Immunology Laboratory at Cincinnati Children's Hospital Medical Center. The Diagnostic Immunology Laboratory provides comprehensive clinical testing to aid in the detection, diagnosis and treatment of pediatric oncologic, hematologic and immunologic disorders.
Dr. Filipovich focused her comments on the importance of pediatric norms in the diagnosis and treatment of patients with rare immune disorders and how staff collaborate with physicians across the country to enhance pediatric patient care.

## How has the field of diagnostic immunology changed in the last decade?

The field of diagnostic immunology has advanced significantly over the years. As technology improves, we are able to provide accurate results in shorter timeframes with less inconvenience for the patient. We have an active research program at Cincinnati Children's and keeping up with new scientific discoveries and technology has been a core focus for us. As a result, we have identified and developed tests to facilitate the diagnosis of more than 80 immune deficiencies. Our laboratory is particularly suited to the diagnosis of congenital and prematurely lethal immune deficiencies.

## Why are pediatric norms important?

Pediatric norms give you a very reliable quantitative indication of whether a child's test results are normal or abnormal.

Immunologic systems develop over the first 10 years of a child's life, so a healthy adult's baseline may be (and is often) very different from a child's. Most laboratories utilize normal ranges developed with healthy adults; this can be misleading when reporting test results for children, as norms may be very different.

## What is important to consider when seeking a diagnosis for a suspected immune deficiency?

A couple of factors are important to consider. First, it's very important to consider the diagnostic capabilities of the facility. Using a lab that offers the broadest range of testing for children gives physicians a higher likelihood of finding a diagnosis without putting the patient through further testing later. At our lab, we're often able to provide comprehensive diagnosis with just one blood draw.

Cincinnati Children's is proud to be a member of the Jeffrey Modell Centers Network (JMCN).

Another consideration is how much counseling the lab and the physician staff provide to aid with interpretation of results and patient management. Because these conditions are so rare and complex, pediatricians don't deal with them on a regular basis and often need advice from an immunological expert. We provide not only the test results but interpretation of those results and expert advice on next steps.

## What is the benefit of having researchers, diagnostic capabilities and clinical application in the same facility?

Finding a healthy balance between clinical and bench-related research is essential to the continuation of discovery and is more easily done with all three disciplines collaborating in the effort. It's important to not only have the cutting edge science but to connect the science with what is happening in the clinics in a timely manner.

## Can you give an example of a newly-developed test that has changed the landscape of immunological testing?

We have developed a new series of rapid screening tests to identify the majority of prematurely lethal immune deficiencies, allowing treatment to begin sooner. These tests have been developed for disorders where we can identify the defective protein. This allows abnormal rapid screening tests to be confirmed with the direct genetic sequencing confirming the diagnosis.

## Can you explain the importance of having a diagnostics lab integrated with confirmatory genetic testing?

Cincinnati Children's is ranked in the top five for Cancer Care by *U.S. News & World Report* and is one of only 10 pediatric hospitals in the United States included on the Honor Roll in *U.S. News'* 2009 edition of *America's Best Children's Hospitals.*

Incorporating a diagnostics lab with confirmatory genetic testing leads to the most efficient and definitive diagnosis.

For example, at Cincinnati Children's, if a test is abnormal and we have residual sample material from the patient remaining from the first round of tests, we can run further genetic tests without delay and can help families have answers more quickly. Our flow of work is organized in such a way that allows us to go to this next step.

## What does being a FOCIS Center of Excellence mean to your lab?

Being a FOCIS Center of Excellence allows us to open our lab as a training facility to researchers and clinicians, as well as to provide a setting in which to do innovative and advanced work. The complexity of our discipline means there are a limited number of facilities where such multidisciplinary scientific and clinical education can take place. FOCIS Centers of Excellence provide opportunities for new discoveries that otherwise may not take place.

## Call for a Consult

Cincinnati Children's offers the most comprehensive pediatric diagnostic immune deficiencies lab in North America, backed by immunological experts who are available to consult on test results.

For a consultation, call 513-803-2603 or email Linda Carl at linda.carl@cchmc.org.

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