Families get together in struggle against rare disorder

By Scott O'Connell/Gatehouse News Service
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SHREWSBURY — In a downstairs room of the Hoagland-Pincus Conference Center in Shrewsbury on Saturday, about a dozen kids who appear as normal and healthy as any other, played games, watched a magic show and had a ball.

But as Westborough resident Jason Rastad noted, it was a rare opportunity for the kids to feel normal as well. Rastad's son, Alec, like many other kids attending the Immune Deficiency Foundation's annual conference that day, suffers from primary immune deficiency disorder.

"When we moved here, Alec didn't know anyone else (with the disorder)," Rastad said. "Here, the kids get to interact, so they see they're not the only one ... that they're not alone."

Immunodeficiency disorders inhibit the victim's immune system, making him or her much more vulnerable to infectious diseases. While acquired immunodeficiency - especially acquired immune deficiency syndrome, or AIDS - is well known, there is another population of people, like Alec, who are simply born with a compromised immune system.

Doctors usually identify those with primary immune deficiency sufferers in their childhood years, but a lack of information creates a huge gap between the onset of symptoms and the eventual diagnosis - 9.2 years on average, said Jason's wife, Towma.

"My son's had everything," she said. "Diseases that people don't usually get - chronic infections. That's why the 9.2 years is truly unacceptable."

Kids like Alec have to receive regular plasma infusions their entire lives to combat the disorder, a burden that can affect them in many ways.
"He struggles so hard to be normal," Towma Rastad said, who added that Alec sometimes gets frustrated that he is the only one among his friends to have to go through the experience.

"You can't judge a book by its cover. Even though he looks good on the outside, as a parent, it makes you a little bit more protective."

That's why, in addition to giving the kids a chance to meet peers with primary immunodeficiency, Saturday's conference was a chance for their parents to interact with two of the region's leading experts in the field: Dr. Francisco Bonilla and Dr. Manish Butte. The doctors gave talks on immune deficiency throughout the day and fielded questions from parents.

"If you don't educate yourself, how are you going to treat yourself?" Towma said. "We're our own advocates in health care."

Making others aware of immunodeficiency's often misunderstood effects is equally challenging, she said. In addition to the public, Towma said many primary care doctors and nurses often don't have enough information about it.

Doctors in particular must become "lumpers," she said. To identify primary immune deficiency cases, doctors must lump symptoms together to get a whole picture, instead of looking at each symptom separately for a cause.

"If you educate the doctors and nurses and the public in general, that's when you're going to get answers," she said. "You raise more money, you get more research and you get a cure eventually. The hope is there."

For now, however, families like the Rastads must be their own advocates. Towma, who helped organize Saturday's conference, said the day was a welcome change from dealing with their son's disease on their own.

"The first five years (after Alec was born) I kept pinching myself, because I thought it was a bad dream - it's overwhelming," she said, "It's such a rare disease, you may not meet anyone else with it."

That wasn't the case for Alec on Saturday.

"Even if it's just one day of fun with no needles, it's worth it," Towma said.

For more information on primary immune deficiency, visit the Immune Deficiency Foundation's Web site, www.primaryimmune.org.