

THE DAILY CHECKUP BY KATIE CHARLES

When science grants immunity

Problems in the body's defenses can be hard to spot; bone-marrow & stem-cell transplants are among treatments

► The specialist: Dr. Charlotte Cunningham-Rundles on immune deficiency disorders

The director of the Immunodeficiency Clinic at Mount Sinai Medical Center, Cunningham-Rundles specializes in allergy and immunology.

► Who's at risk

As the body's defense mechanism against disease, the immune system identifies and kills menaces like pathogens and tumor cells — but in some people, that system breaks down. “With immune deficiency disorders, your immune system doesn't have all the equipment it needs,” says Cunningham-Rundles. “You don't have all the capacity to fight infection.”

Some relatively mild immune deficiency disorders are fairly common, while the odds of having a more serious variety are 1 in 10,000.

The disorders fall into two main groups. “Primary immune deficiency means the disease is genetic, and that is the way your system was created,” says Cunningham-Rundles. “Secondary immune deficiency means that your immune system is breaking down because of something in the environment, like chemo, steroids or HIV.” While primary immune deficiency runs in some families, in others, it occurs randomly.

As for gender: “Some disorders only run in boys because they are gene defects on the X chromosome, and girls have the backup of the other X,” says Cunningham-Rundles. “But globally, these disorders are equal in affecting men and women.”

Immune deficiency disorders are present in all populations. “No one is at more at risk than anyone else,” says Cunningham-Rundles. Doctors have identified 150 types of immune deficiency disorders, and they continue to discover a few new ones each year. Some types are mild, and others are far more severe.

Many people are familiar with the idea of immune deficiency from the story of “the boy in the bubble.” “In that type, severe combined immune deficiency, the immune system has a block on making certain cells, so the disorder becomes obvious when the baby is very young, probably in the first month of life,” says Cunningham-Rundles. “In contrast, with some of the mild forms, you might not discover the disease until you're 30 or 40, since the body has found ways to compensate.”

► What you can do

Get informed.

The Immune Deficiency Foundation (primaryimmune.org) has a “patients and families” section with information on insurance, clinical trials and peer support. The Jeffrey Modell Foundation has lists of warning signs and a search engine for specialists (info4pi.org).

Keep your immune system strong.

Everyday things like exercise, sticking to a healthy lifestyle and not smoking can help boost your immunity. “Doing these things means your system isn't exposed to extra unhealthy particles,” says Dr. Cunningham-Rundles.

Pay attention to infections.

Many patients go undiagnosed for years, while suffering through a series of severe infections. “A couple of bouts of pneumonia that were proven by chest X-ray, or being hospitalized with a bloodstream infection, are big warning signs of immune deficiency,” says Cunningham-Rundles.



Genetic research is shedding more and more light on immune disorders, says Dr. Charlotte Cunningham-Rundles.

► Signs and symptoms

Immune deficiency disorders can be easier to diagnose in children, who tend to show more obvious symptoms than adults. “Pediatricians look out for rash, failure to thrive, diarrhea, anemia and recurring pneumonias, especially as the result of an opportunistic organism,” says Cunningham-Rundles.

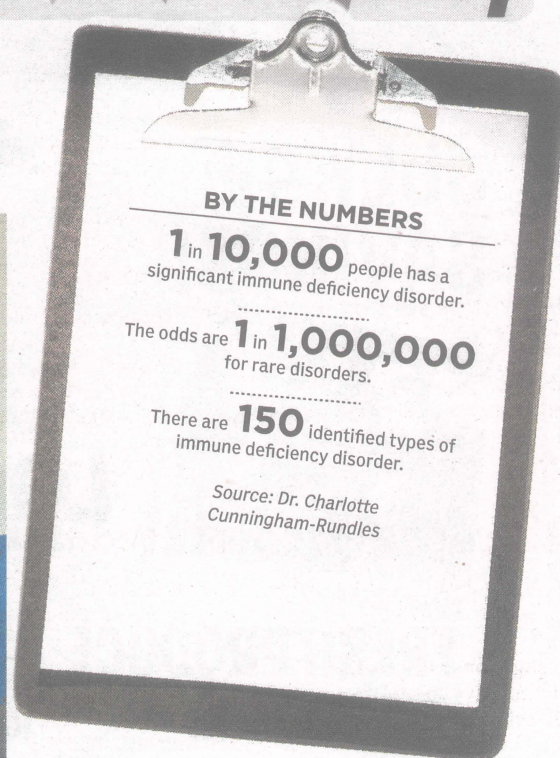
Adults can look pretty healthy despite their immune deficiency disorder and often go years before they're diagnosed. “The most common symptom is a pattern of frequent respiratory problems — sinusitis, bronchitis, pneumonia,” says Cunningham-Rundles. “For people with an immune deficiency disorder, they probably will end up in the hospital as the result of the pneumonia.”

Suffering from multiple pneumonias that have been diagnosed by chest X-ray, or blood infections that lead to hospitalization, are red flags for immune deficiency.

Many internists aren't trained to spot immune deficiency disorders, and it's harder for a patient visiting lots of specialists to have someone pick up on the pattern of illnesses. By the time they reach an expert, most patients are usually pretty frustrated — and relieved once they are diagnosed. “They feel like some part of the puzzle has been put together,” says Cunningham-Rundles. “They finally understand why they had recurrent infections throughout their life.”

► Traditional treatment

The options depend on the type of disorder and its severity. “Lots and lots of antibiotics” are usually part of the treat-



ment. “For people who lack antibody, we can give injections of intravenous or subcutaneous gamma globulin, which is the antibody that's been extracted from human blood,” says Cunningham-Rundles. “Depending on the root and the dose, you get shots as frequently as once a week and as little as once a month.” These shots are given for the rest of your life. “It has great results,” says Cunningham-Rundles.

More severe defects call for a transplant of either bone marrow or stem cells. Bone-marrow transplants require finding a good match, usually a relative, and then extracting marrow. Stem cells now often come from a stem-cell bank that stores placental blood.

Once there's a match, the recipient usually needs chemotherapy to get his or her bone marrow to make space for the transplant. “In the transplant, we insert the extracted marrow or stem cells through the veins,” says Cunningham-Rundles. “The transplant finds its way to the bone marrow and takes up residence there.”

Although this treatment can't cure the immune deficiency disorder completely, it can improve quality of life and life span. “When it works, it's wonderful,” says Cunningham-Rundles. “Some of our patients who might not have survived before are now 25 or 30 and in medical school.”

► Research breakthroughs

Doctors' understanding of immune deficiency disorders is growing by leaps and bounds as genetic research becomes more sophisticated. “We keep discovering different defects, two or three a year for the past few years,” says Cunningham-Rundles. “When you have a patient with a newly discovered gene defect, that casts light on a lot of other problems.” In many cases, researchers have identified the gene that causes the defect, and doctors can use that information to personalize the treatment.

► Questions for your doctor

If you've had recurrent significant infections, ask your physicians, “What's going on with my immune system?” And don't be shy about asking, “Can you refer me to an immunologist?” Not all doctors have been trained to spot immune deficiency disorders. “The awareness of these diseases is an issue all around,” says Cunningham-Rundles.

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