World PI Week

PI Week Leaflet

What is PI ?	When a defect in the immune system is inherited (carried through the genes), it is called primary, or inherited, immune deficiency. There are over 150 forms of Primary Immunodeficiency (PI), ranging widely in severity. PI often presents itself in the form of "ordinary" infections. Physicians sometimes treat the infections while missing the underlying cause, allowing the illnesses to recur, and leaving the patient vulnerable to vital organ damage, physical disability, and even death. Families struggling with immunodeficiency often do not know where to turn for help. Social, emotional, and financial burdens can often be overwhelming. The problems presented in inherited immunodeficiency disease have challenged researchers and immunologists to reach improved diagnoses, treatments, and new therapies. Promising results in this area of immunology are also yielding benefits for victims of cancer, AIDS, autoimmunity, cystic fibrosis, and a wide range of pulmonary and allergic conditions.
What are the symptoms?	There are a number of different signs and symptoms of a Primary Immunodeficiency disease. Although it varies somewhat from individual to individual and disorder to disorder, there are some signs and symptoms that occur commonly in many of the disorders. Perhaps the most common problem that patients with a PI disease have is increased susceptibility to infection. PI patients have too many, and often recurrent, infections. They may have recurrent and repeated ear infections, pneumonia, bronchitis, sinusitis or skin infections. Although less common, PI patients may have abscesses of their internal organs, such as the liver, or infections of their blood; however, the common theme is that they have more infections than they or their doctor believes is appropriate. Not every patient with a PI disease demonstrates recurring infections. In some patients, the first infection is serious enough to render the possibility of a PI disease. There are certain infections caused by germs that only afflict PI patients, therefore the type of infection itself may serve as a red flag for PI disease. Patients with PI diseases may also present with a variety of autoimmune or rheumatologic problems. In some cases, the rheumatic disorder can take the form of systemic lupus erythematosis or rheumatoid arthritis and involve many organs and tissues. However, the rheumatic disorder may also only affect one organ and take the form of an isolated arthritis, kidney disease, thyroid disease, low platelet counts in the blood, or anemia. Furthermore, gastrointestinal (digestive) problems may occur in some patients with a Primary Immunodeficiency disease. Patients may have excessive cramping, loss of appetite, nausea and diarrhea. In some patients, the gastrointestinal problems can be the result of an infection of the intestines - in others, they may be a reflection of an autoimmune or rheumatic disorder

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Who can I consult to get diagnosis?	Correct diagnosis of a PI disease begins with awareness of the 10 Warning Signs and the first step in diagnosing a Primary Immunodeficiency disease is a good evaluation. An immune system specialist (immunologist) can help with diagnosis and treatment. Evaluation of the immune system may include: Detailed medical history Physical exam Blood tests Vaccines to test the immune response At the time of the evaluation, your doctor will ask questions about your health. Frequent or unusual infections, prolonged diarrhea, and poor childhood growth are some symptoms of a possible PI. Because some PI diseases run in families, you may also be asked questions about your family history. If a PI disease is suspected, a series of blood tests and vaccines may be required. Blood tests will show if any part of the immune system is missing or not working properly. Vaccines may be given to test the immune system's response, i.e. its ability to fight invaders.
What treatment can I get?	Once a diagnosis is established, much can be done for PI patients. At a minimum, the recurring infections can be treated with low or moderate doses of appropriate antibiotics. These can help prevent permanent damage to the lungs and bronchial tubes, thus promoting the patient's long-term survival while improving the quality of life. When appropriate, immunoglobulin therapy is the accepted protocol for a wide range of PI diseases. Advanced treatments such as the interleukins, PEG-ADA, and gamma interferon can help in some complex cases. Bone marrow transplantation and gene therapy may be the appropriate protocol in specific disorders.
Is nutrition important?	Good nutrition gives the body the energy and the resources to fight infections. Eating right always makes good sense, and families with antibody deficiencies should take extra care to maintain a healthy and balanced diet.
Can I lead a normal life?	Thanks to new therapies, greater public awareness, and better access to information, many patients with PI are leading more normal lives - going to school, camp, work, playing sports, and enjoying a better quality of life. There has never been more hope for people who are immunodeficient.

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