

Talking Points on Primary Immunodeficiency Diseases

About Primary Immunodeficiency Diseases (PI)

- Primary immunodeficiency diseases (PI) occur in individuals in which part of the immune system is missing or functions improperly. The International Union of Immunological Societies (IUIS) recognizes more than 350 different types of PI, all caused by genetic or intrinsic defects.
- In the U.S., there are approximately 250,000 people diagnosed with PI and thousands more go undetected. This means that in the U.S., approximately 1 in 1,200 persons have a diagnosed PI.
- People with PI live their entire lives more susceptible to infections, and without proper treatment may endure recurrent health problems, often developing serious and debilitating illnesses.
- While not contagious, primary immunodeficiency diseases are caused by hereditary or genetic defects.
- Although some disorders present at birth or in early childhood, the disorders can affect anyone, regardless of age or gender.
- Early diagnosis of PI is critical:
 - Based on Immune Deficiency Foundation (IDF) survey data, the average time from symptom onset to diagnoses is between 9 and 15 years.
 - During this period of time, 37% of patients report permanent functional impairment, including lung disease. With earlier diagnosis, many of these permanent impairments could be avoided.

Countering Misperceptions

- Having PI does not mean you have to live in a bubble.
- While PI is serious, these diseases vary widely in their severity.
- David Vetter, known as the "boy in the bubble" had Severe Combined Immune Deficiency (SCID), the most serious disorder. The only treatment for SCID is a bone marrow transplant, but for some forms of SCID, there have been some promising signs of success from gene therapy.
- With early diagnosis and proper treatment therapy, most patients with PI can go to school, work and live healthy, productive lives.

How do you know if you have a PI?

IDF and its Medical Advisory Committee have developed a list of five key factors to consider if you are suspicious that you or a loved one might have PI. You should be suspicious if you have an infection that is:

Severe – requires hospitalization or intravenous antibiotics

Persistent – won't completely clear up or clears slowly

Unusual – caused by an uncommon organism

Recurrent – keeps coming back

Runs in the Family – others in your family have a similar susceptibility to infection

If any of these describe your infection, ask your physician to check for the possibility of a PI. People with PI are more susceptible to infections and health problems that lead to serious and debilitating diseases. It is critical to get an early diagnosis and proper medical care.