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Primary Immune Deficiency Fact Sheet

What It Is

Primary immune deficiencies (PIDs) are a group of more than 200 diseases that affect the cells, tissues and proteins of the immune system.[1,2] In people with PID, the immune system is either absent or functioning inadequately, leaving them more susceptible to infection. Some PIDs affect a single cell in the immune system while others may affect one or more functions of the system.[2]

Causes and Symptoms

Most PIDs are inherited diseases that are passed down from parent to child. In these cases, a family history is usually present. Others are due to an unknown cause, but generally believed to be related to both genetic and environmental factors.[3]

Symptoms of PID vary and depend on the deficiency of the immune system. The most common symptom of PID is recurrent or persistent infections. Infections may be of the skin, sinuses, throat, ears, lungs, brain or spinal cord, or urinary or intestinal tracts.[2] Additional symptoms include:

- Severe infections requiring hospitalization
- Unusual infections caused by an uncommon organism
- Chronic sinus and/or ear infections
- Recurrent pneumonia[4]
- Chronic diarrhea[1]
- Need for intravenous antibiotics to clear infection because regular oral antibiotics fail
- A history of infection susceptibility in the family[4]
- Failure of infant to gain weight or grow normally[4]

Incidence and Prevalence

Originally thought to be extremely rare, certain variations of PID have become relatively common. For example, Selective IgA deficiency occurs in as many at 1 in 500 to 1 in 1000 individuals. Some of the other less common PIDs may only occur in 1 in 10,000 to 1 in 100,000 individuals. In Canada there are more than 2,000 people diagnosed with PIDs, although the condition is thought to be widely undiagnosed.[1]

PIDs can occur in individuals of any age or sex.[2] While primarily diagnosed in children, PIDs have now been shown to occur in adolescents and adults.[1]

Living with PID

Living with PID can be both frightening and frustrating as the illnesses associated with the condition can be quite serious and the lack of a definite diagnosis leaves many unanswered questions. For parents of school-age children, a general lack of awareness among the public, including teachers and school nurses, can cause increased anxiety.[5]

Diagnosis and Treatment

Early detection is critical to limit any permanent damage caused by severe infections.[1]

PID is diagnosed by studying the nature of the infections, as well as the additional symptoms of the sickness. A definitive diagnosis is conducted through immunologic testing, which includes a full blood count and a measure of immunoglobulin levels.

Treatment for PID depends on the nature of the abnormality. Recurrent infections can be treated with appropriate antibiotics. When appropriate, immunoglobulin replacement therapy is considered the standard treatment for a wide range of PIDs. 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 appropriate for specific disorders.[5]

References

[1] Canadian Immunodeficiencies Patient Organization brochure. Accessed 4/30/09: http://www.cipo.ca/cipo%20pamhlet%20good%20copy.pdf

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