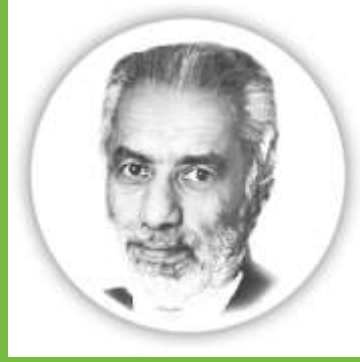

بسم الله الرحمن الرحیم

دانشگاه علوم پزشکی
و خدمات بهداشتی درمانی تهران



Patient Education in Primary Immunodeficiency

(۱۳۷۵)
انجمن آسم و آلرژی ایران
IRANIAN SOCIETY OF ASTHMA & ALLERGY
(ISAA)

بیمارستان کودکان حکیم
Hakim Children Hospital

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IRANIAN SOCIETY OF PEDIATRICS

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What is PI?

Primary immunodeficiencies(PIs) are agroup ofmore than 550 rare, chronic conditions where a part of the body's immune system is missing or does not function correctly All primary immunodeficiencies are different, but they all disrupt the body's immune system.

They are also known as inborn errors of immunity (IEI).

What are the signs of a primary immunodeficiency?

PI conditions prevent the immune system from working as it should, so the body has a hard time protecting itself against infection.

This increased susceptibility can lead to infections that are:

- **Recurrent (keep coming back).**
- **Persistent (won't completely clear up or clear very slowly).**
- **Unusual (caused by an uncommon organism).**
- **Severe (require hospitalization or intravenous antibiotics).**
- **Shared by family members (others in your family have or have had a similar susceptibility to infection).**

Keep in mind that infections can occur anywhere in the body, such as the brain or spinal cord, ears, lungs, skin, sinuses, throat, and urinary or intestinal tracts.

Additional symptoms of PIs include:

Swollen spleen, liver, or lymph nodes (splenomegaly, hepatomegaly, or lymphadenopathy).

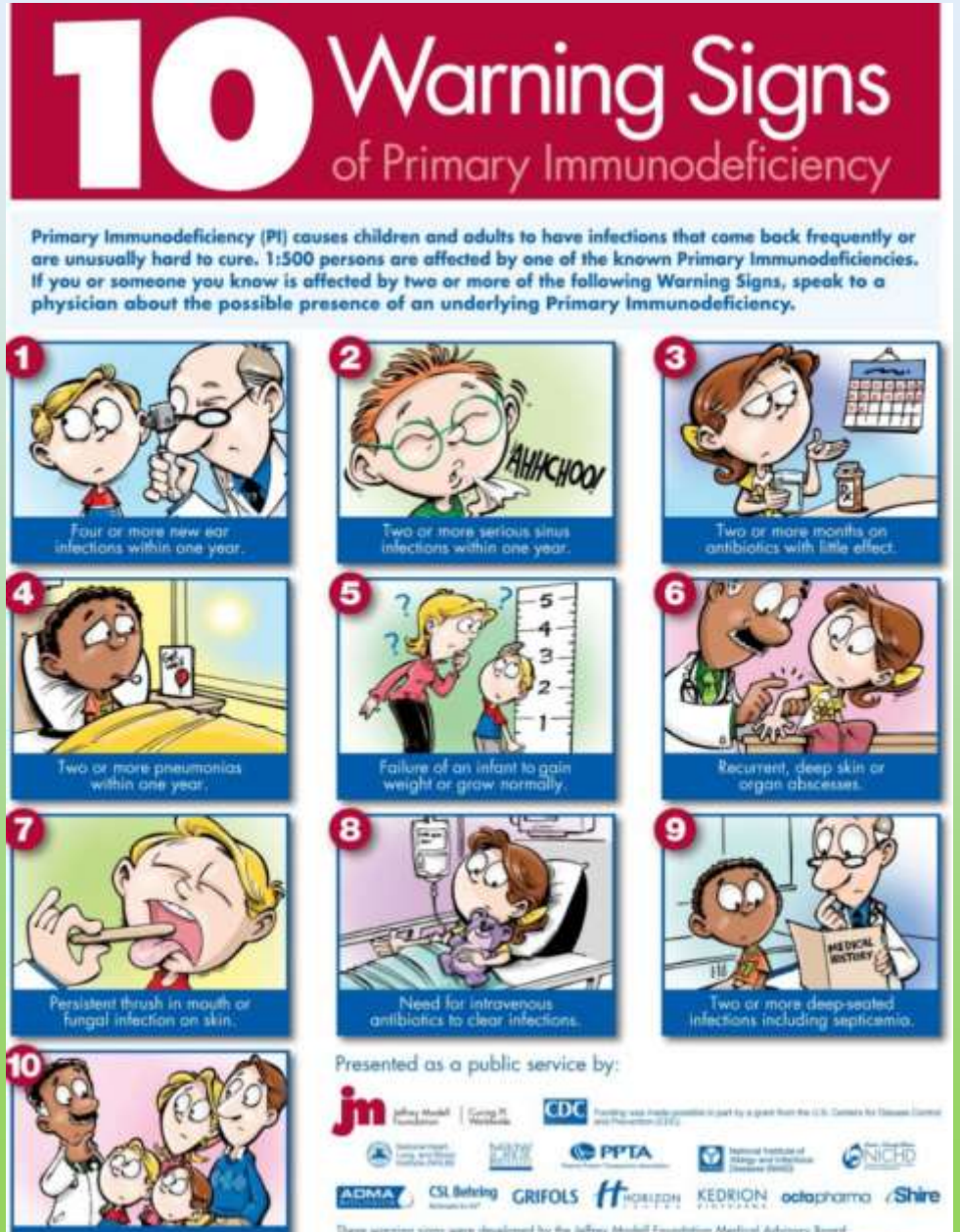
Inflammation of blood vessels (vasculitis).

Autoimmune or autoinflammatory symptoms like inflammatory bowel disease.

If any of these phrases describe your symptoms, ask your doctor to check for the possibility of a primary immunodeficiency (PI).

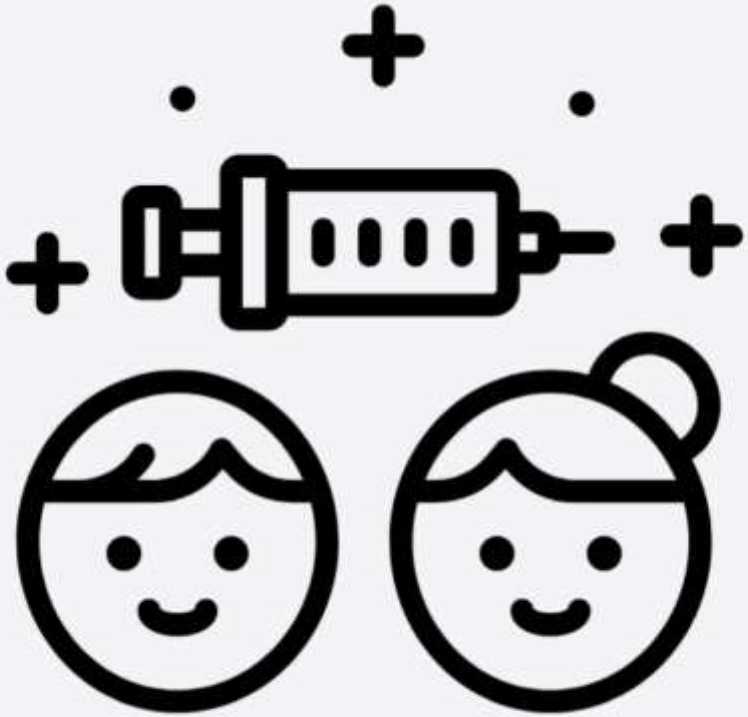
Who is at risk for PI?

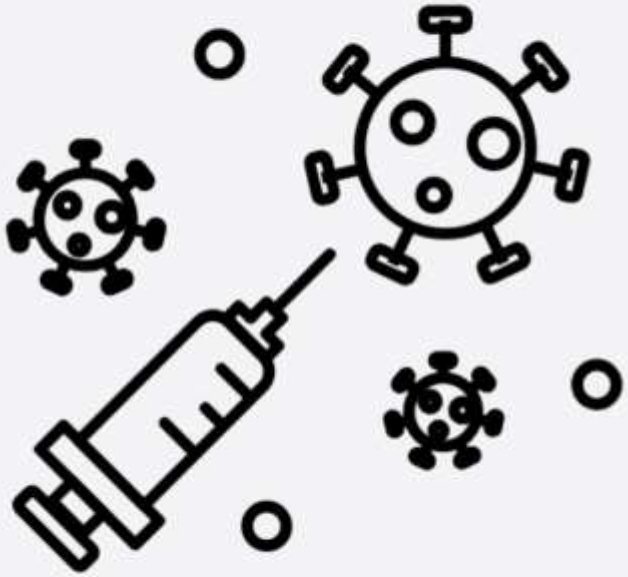
PI conditions are caused by genetic variants and can affect anyone, regardless of age, gender, or ethnicity. Some PI disorders cause symptoms in infancy or early childhood, but others may not present until mid- or even older adulthood.



1. Breastfeeding

- **General Recommendation:** Breastfeeding is generally encouraged for infants with PI due to the immunological benefits conferred by maternal antibodies, particularly secretory IgA.
- **Exceptions:** In cases where the mother is seropositive for cytomegalovirus (CMV) and the infant has severe combined immunodeficiency (SCID), breastfeeding may be contraindicated due to the risk of CMV transmission.
- **Clinical Guidance:** Each case should be evaluated individually, considering the type of immunodeficiency and maternal CMV status.





2. Vaccination

- **Inactivated Vaccines:** Generally safe and recommended for PI patients. However, the immunogenic response may be suboptimal, necessitating monitoring of antibody titers.
- **Live Attenuated Vaccines:** Typically contraindicated in patients with significant T-cell deficiencies (e.g., SCID, Wiskott-Aldrich syndrome) due to the risk of vaccine-derived infections.
- **Household Contacts:** Should be fully vaccinated to provide herd immunity, especially with annual influenza vaccines and COVID-19 vaccines.

3. Infection Prevention

- **Hygiene Practices:** Emphasize regular handwashing with soap and water, respiratory etiquette, and routine dental care.
- **Environmental Exposure:** Advise avoidance of crowded places during peak infection seasons and close contact with individuals exhibiting signs of infection.
- **Prophylactic Therapies:** Consider prophylactic antibiotics, antifungals, or antivirals based on the specific PI diagnosis and history of infections.
- **Immunoglobulin Replacement:** For patients with significant antibody deficiencies, regular intravenous or subcutaneous immunoglobulin therapy is essential.

4. Social Interactions and Public Spaces

- **Daycare and School Attendance:** Decisions should be individualized based on the severity of the immunodeficiency, current treatment regimen, and infection control measures in place.
- **Social Activities:** Encourage participation in social activities with appropriate precautions to promote psychosocial well-being.
- **Education of Peers and Staff:** Provide information to educators and caregivers about the child's condition, necessary precautions, and emergency protocols.

5. Surgical and Invasive Procedures

- **Preoperative Assessment:** Thorough evaluation of immune status, including recent infection history and current immunoglobulin levels.
- **Antibiotic Prophylaxis:** May be indicated depending on the type of PI and the nature of the surgical procedure.
- **Postoperative Care:** Close monitoring for signs of infection and prompt management of any complications.

6. Dietary Considerations

- **Balanced Nutrition:** Encourage a well-balanced diet rich in fruits, vegetables, whole grains, lean proteins, and healthy fats to support overall health.
- **Food Safety:** Advise avoidance of raw or undercooked meats, unpasteurized dairy products, and other high-risk foods to minimize exposure to foodborne pathogens.
- **Nutritional Supplements:** Supplementation should be based on documented deficiencies; indiscriminate use is discouraged.

7. Hygiene and Personal Care

- **Skin Care:** Maintain regular bathing routines using mild soaps; promptly address any skin lesions or infections.
- **Oral Hygiene:** Encourage regular brushing, flossing, and dental check-ups to prevent oral infections.
- **Hair and Nail Care:** Use personal grooming tools; avoid communal instruments in salons to reduce infection risk.

8. Medication and Supplement Use

- **Medication Adherence:** Emphasize the importance of adhering to prescribed treatment regimens, including immunoglobulin therapy and prophylactic medications.
- **Over-the-Counter Products:** Advise consultation with healthcare providers before initiating any new medications or supplements, including herbal remedies.
- **Probiotics:** Generally not recommended for patients with significant immunodeficiencies due to the risk of systemic infections.

9. Mental Health and Psychosocial Support

- **Psychological Well-being:** Recognize the emotional impact of chronic illness; provide resources for counseling and support groups.
- **Family Support:** Educate family members about the condition to foster a supportive home environment.
- **Transition of Care:** Prepare adolescents for the transition to adult care services through structured programs.

Traveling safely

People with primary immunodeficiency (PI) don't have to stay home—it may take a bit of extra planning, but you can get out and see the world, just like anyone else. However, taking your PI into consideration while planning your trip is crucial to safe, healthy, and enjoyable travel.

Where are you going?

Some places and activities are riskier for a person with PI. For example, some immunologists tell people with chronic granulomatous disease (CGD) not to swim in the ocean or fresh water. Likewise, people who do not respond to vaccines shouldn't visit places where they are likely to be around hard-to-treat germs.

Don’t leave your medications in a hot car.

I Am Immunocompromised emergency card

Are you flying?

The air on most planes is HEPA filtered so flying is generally safe, even without a mask. However, you can wear a KN95 or N95 mask for added protection and peace of mind.

If you are flying, take your medications in your carry-on luggage; it's generally too cold in the baggage compartment.

Do the countries you're visiting require proof of vaccination?

If you don't get vaccines because you don't respond to them, you will need a letter from your immunologist explaining why you are not vaccinated.

What are the rules for bringing prescription drugs or medical supplies/equipment with you?

Keep medications in their original containers with the prescription label and let security know you have medication or supplies that may need to be screened.

Ask your healthcare provider to write a letter of necessity for all medications and medical equipment and carry it with you. This letter is especially important if you are carrying on liquid medications, needles or other sharps, or will be going through customs.

Make sure there is an appropriate place to store your medication where you are staying.

For example, you may need to request a hotel room with air conditioning or a refrigerator if you are traveling to a hot place.

Autosomal recessive inheritance involves gene variants located on the 22 numbered chromosome pairs (autosomes). For a person to be affected by an autosomal recessive primary immunodeficiency (PI), they must inherit two copies of the disease-causing gene variant—one from each parent. Typically, parents are carriers, each with one faulty and one normal gene copy, and they do not show symptoms.

When both parents are carriers:

- **Each child has a 25% chance of being affected,**
- **A 50% chance of being a carrier,**
- **And a 25% chance of inheriting no faulty gene.**

These probabilities remain the same for every pregnancy, regardless of past outcomes.

Examples of autosomal recessive PIs include:

- **Certain types of Severe Combined Immunodeficiency (SCID), such as ADA, RAG, JAK3, and Artemis SCID,**
- **MHC class II deficiency,**
- **Some types of Chronic Granulomatous Disease (CGD),**
- **Leukocyte Adhesion Deficiency (LAD),**
- **Chediak-Higashi syndrome,**
- **Familial Hemophagocytic Lymphohistiocytosis (HLH).**

These conditions are rare, often with no family history, but are more common in consanguineous marriages or certain populations with higher carrier frequencies.

In autosomal dominant inheritance, only one copy of a disease-causing gene variant is needed for a person to develop the condition, regardless of the other gene copy.

Both males and females can be affected. If a parent has the condition, each child has a 50% chance of inheriting it, and this risk remains the same for every pregnancy.

Sometimes, the gene variant appears for the first time in a family due to a new (spontaneous) mutation in the fertilized egg.

Examples of autosomal dominant primary immunodeficiencies (PIs) include:

- **Hyper IgE syndrome (Job's syndrome, due to STAT3 loss of function),**
- **WHIM syndrome (warts, hypogammaglobulinemia, infections, and myelokathexis),**
- **Certain rare defects in the IFN-γ/IL-12 pathway,**
- **STAT1 and STAT3 gain of function immune disorders.**

X-linked recessive inheritance involves gene variants located on the X chromosome. Males, having only one X chromosome, are affected if that chromosome carries the disease-causing variant, as they lack a second X to compensate. Females have two X chromosomes and are typically carriers if they inherit one faulty copy, as the second (normal) copy usually maintains function.

Inheritance patterns:

- Affected males pass the variant to all daughters (who become carriers) but none of their sons (who inherit their Y chromosome).
- Carrier females have a 50% chance of passing the variant to each child:
    - 25% chance of having an affected son,
    - 25% chance of an unaffected son,
    - 25% chance of an unaffected, non-carrier daughter,
    - 25% chance of a carrier daughter.

This probability remains the same in each pregnancy.

Some female carriers may show symptoms due to X-inactivation, a random process during development where one X chromosome is silenced in each cell. If the healthy X is inactivated in many cells, the faulty X may cause symptoms.

Spontaneous mutations can also cause X-linked PIs in individuals with no family history.

Examples of X-linked recessive primary immunodeficiencies (PIs):

- **X-linked agammaglobulinemia (XLA),**
- **Wiskott-Aldrich syndrome,**
- **SCID (common gamma chain variants),**
- **Hyper IgM syndrome (CD40 ligand variants),**
- **X-linked lymphoproliferative disease (2 forms),**
- **Most common form of chronic granulomatous disease (CGD),**
- **Properdin deficiency,**
- **Dyskeratosis congenita.**

10 warning signs of Primary Immunodeficiency

Primary Immunodeficiency (PI) causes children and adults to have infections that come back frequently or are unusually hard to cure. 1:500 persons are affected by one of the known Primary Immunodeficiencies. If you or someone you know is affected by two or more of the following Warning Signs, speak to a physician about the possible presence of an underlying Primary Immunodeficiency.

Four or more new ear infections within one year

Two or more serious sinus infections within one year

Two or more months on antibiotics with little effect

Two or more pneumonias within one year

Failure of an infant to gain weight or grow normally

Recurrent, deep skin or organ abscesses

Persistent thrush in mouth or fungal infection on skin

Need for intravenous antibiotics to clear infections

Two or more deep-seated infections including septicemia

A family history of PI

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MALAYSIAN PATIENT ORGANISATION FOR PRIMARY IMMUNODEFICIENCIES (MYPOPI)

MyPOPI is a non-profit organisation and registered society caring for patients and supporting families affected by Primary Immunodeficiencies (PID)

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References

- UpToDate, Immune Deficiency Foundation, Mayo Clinic, National Jewish Health, StatPearls.


THANK YOU