Immune thrombocytopenia

Immune thrombocytopenia is a disorder characterized by a blood abnormality called thrombocytopenia, which is a shortage of blood cell fragments called platelets that are needed for normal blood clotting.

Affected individuals can develop red or purple spots on the skin caused by bleeding just under the skin's surface. Small spots of bleeding under the skin are called purpura and larger spots are called ecchymoses. People with immune thrombocytopenia can have significant bleeding episodes, such as nose bleeds (epistaxis) or bleeding in the moist lining (mucosae) of the mouth. In severe cases, individuals may have gastrointestinal bleeding or blood in the urine or stool, or heavy and prolonged menstrual bleeding (menorrhagia). In very rare instances, bleeding inside the skull (intracranial hemorrhage) can occur, which can be life-threatening. A greater reduction in platelet numbers is often associated with more frequent bleeding episodes and an increased risk of severe bleeding.

While immune thrombocytopenia can be diagnosed at any age, there are two periods when the condition is most likely to develop: early childhood and late adulthood. In children, the reduction in platelets is often sudden, but platelet levels usually return to normal levels within weeks to months. Immune thrombocytopenia in children is often preceded by a minor infection, such as an upper respiratory infection, but the relationship between the infection and immune thrombocytopenia is not clear. In adults, the development of immune thrombocytopenia is usually gradual and the condition tends to persist throughout life.

Frequency

The incidence of immune thrombocytopenia is approximately 4 per 100,000 children and 3 per 100,000 adults. In adults with immune thrombocytopenia, women are affected more often than men.

It is likely that this condition is underdiagnosed because those with mild signs and symptoms often do not seek medical attention.

Causes

The genetic cause of immune thrombocytopenia is unclear. This condition occurs when the body’s own immune system malfunctions and attacks the body's tissues and organs (autoimmunity). Normally, the immune system produces proteins called antibodies, which attach to specific foreign particles and germs, marking them for destruction. In immune thrombocytopenia, the immune system abnormally destroys platelets and makes fewer platelets than normal. People with immune thrombocytopenia produce antibodies that attack normal platelets. The platelets are destroyed and eliminated from
the body, resulting in a shortage of these cells in affected individuals. Some of these antibodies also affect the cells in the bone marrow that produce platelets (known as megakaryocytes), which leads to a decrease in platelet production, further reducing the number of platelets in the blood.

In some people with immune thrombocytopenia, the abnormal immune reactions may coincide with an infection by certain viruses or bacteria. Exposure to these foreign invaders may trigger the body to fight the infection, but the immune system also mistakenly attacks platelets.

Genetic variations (polymorphisms) in a few genes have been found in some people with immune thrombocytopenia and may increase the risk of abnormal immune reactions. However, the contribution of these genetic changes to the development of immune thrombocytopenia is unclear.

When the condition is due to the targeted destruction of platelets by the body's own immune cells, it is known as primary immune thrombocytopenia. Immune thrombocytopenia following bacterial or viral infection is considered primary because the infection triggers a platelet-specific immune reaction, typically without any other signs or symptoms. However, immune thrombocytopenia can be a feature of other immune disorders, such as common variable immune deficiency, which occurs when the immune system has a decreased ability to protect the body against foreign invaders, or other autoimmune disorders such as systemic lupus erythematosus. Immune thrombocytopenia can also occur with other blood disorders, including a form of cancer of the blood-forming tissue known as chronic lymphocytic leukemia, and human immunodeficiency virus (HIV) infection. When immune thrombocytopenia is a feature of other disorders, the condition is known as secondary immune thrombocytopenia.

**Inheritance Pattern**

Immune thrombocytopenia and other autoimmune disorders can run in families, but the inheritance pattern is usually unknown. People with a first-degree relative (such as a parent or sibling) with immune thrombocytopenia likely have an increased risk of developing the disorder themselves.

**Other Names for This Condition**

- autoimmune thrombocytopenia
- autoimmune thrombocytopenic purpura
- idiopathic thrombocytopenic purpura
- immune thrombocytopenic purpura
- ITP
- Werlhof disease
Diagnosis & Management

Genetic Testing Information

• What is genetic testing?
  /primer/testing/genetictesting

• Genetic Testing Registry: Autoimmune thrombocytopenia

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22immune+thrombocytopenia%22+OR+%22idiopathic+thrombocytopenic+purpura%22+OR+%22immune+thrombocytopenic+purpura%22

Other Diagnosis and Management Resources

• Johns Hopkins Medicine
  https://www.hopkinsmedicine.org/health/conditions-and-diseases/idiopathic-thrombocytopenic-purpura

• MedlinePlus Encyclopedia: Idiopathic Thrombocytopenic Purpura (ITP)
  https://medlineplus.gov/ency/article/000535.htm

• Seattle Children’s Hospital
  https://www.seattlechildrens.org/conditions/heart-blood-conditions/itp

Additional Information & Resources

Health Information from MedlinePlus

• Encyclopedia: Idiopathic Thrombocytopenic Purpura (ITP)
  https://medlineplus.gov/ency/article/000535.htm

• Encyclopedia: Purpura
  https://medlineplus.gov/ency/article/003232.htm

• Health Topic: Platelet Disorders
  https://medlineplus.gov/plateletdisorders.html

Genetic and Rare Diseases Information Center

• Idiopathic thrombocytopenic purpura
  https://rarediseases.info.nih.gov/diseases/5194/idiopathic-thrombocytopenic-purpura

Additional NIH Resources

• National Heart, Lung, and Blood Institute
  https://www.nhlbi.nih.gov/health-topics/immune-thrombocytopenia
Educational Resources

- Ann & Robert H. Lurie Children's Hospital of Chicago

- Children's Hospital of Philadelphia
  https://www.chop.edu/conditions-diseases/idiopathic-thrombocytopenic-purpura-itp

- Cincinnati Children’s Hospital
  https://www.cincinnatichildrens.org/health/i/itp

- Dana-Farber Cancer and Blood Disorders Center at Boston Children’s Hospital

- MalaCards: thrombocytopenic purpura, autoimmune
  https://www.malacards.org/card/thrombocytopenic_purpura_autoimmune

- Merck Manual Consumer Version
  https://www.merckmanuals.com/home/blood-disorders/platelet-disorders/immune-thrombocytopenia

- Orphanet: Immune thrombocytopenic purpura
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3002

Patient Support and Advocacy Resources

- Contact a Family (UK)
  https://contact.org.uk/medical-information/conditions/i/immune-thrombocytopenia/

- Foundation for Women & Girls with Blood Disorders
  http://www.fwgbd.org/

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/immune-thrombocytopenia/

- Platelet Disorder Support Association
  https://www.pdsa.org/

- The ITP Support Association (UK)

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Purpura,+Thrombocytopenic,+Idiopathic%5BMAJR%5D%29+AND+%28immune+thrombocytopenia%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- THROMBOCYTOPENIC PURPURA, AUTOIMMUNE
  http://omim.org/entry/188030

Medical Genetics Database from MedGen

- Immune thrombocytopenia

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