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Pediatric Blood Diseases

By Yolanda Smith, BPharm

There are several blood diseases that may affect infants, children and adolescents. These include hemoglobinopathies and bone marrow failure syndromes, as well as the hemophilias. Acquired and auto-immune blood disorders may also occur, including iron-deficiency anemia and idiopathic thrombocytopenic purpura.

Beta-Thalassemia

Beta-thalassemia is usually diagnosed within the first 12 months of birth. It is most common in children of Mediterranean, Asian or African origin. Affected children are unable to produce the globin protein, which is an essential component of hemoglobin. As a result, there is a deficiency in the oxygen supply to the body because the red blood cells lack oxygen-carrying capacity. This causes a range of symptoms such as fatigue and pallor, as well as an enlarged heart, spleen and liver.

Sickle Cell Disease

Sickle cell disease involves the production of abnormal beta-globin molecules within the hemoglobin. This deficiency causes the red blood cells to become sickle-shaped. As a result, the red blood cells cannot move through the blood capillaries as freely as normal. Therefore some body tissues and organs may have an inadequate blood supply.

Fanconi Anemia

Fanconi anemia is the most common type of bone marrow failure. It is usually diagnosed in early childhood. Affected children are unable to produce white and red blood cells and platelets due to one of several inherited gene mutations. It can cause skin pigmentation, abnormal eye size and short stature. These individuals are at a higher risk for leukemia and the development of other tumors.

Dyskeratosis Congenita

Dyskeratosis congenita may be caused by one of several inherited gene mutations, several of which are known. Approximately half the cases are caused by an unknown mutation. It usually presents in adolescence and causes patches on the skin and in the mouth, as well as nail changes. The gene abnormality interferes with the production of white cells, red cells and platelets. It can be diagnosed by a screening test known as telomere length analysis.

Shwachman-Diamond Syndrome

Shwachman-Diamond syndrome is a blood disorder that is usually caused by the inheritance of a mutation in the SBDS gene. It primarily affects the production of white blood cells, but can also affect red blood cells and platelets in advanced cases. It is usually diagnosed in childhood, and causes bone abnormalities and fat malabsorption, due to the impaired function of the liver and pancreas.

Diamond-Blackfan Anemia

Diamond-Blackfan anemia is a bone marrow disorder that affects the production of certain proteins, due to an inherited gene mutation. This usually reduces the production of red blood cells. White cell and platelet production remains relatively constant. Affected children may have other physical abnormalities, such as odd-shaped thumbs and short stature.

Congenital Amegakaryocytic Thrombocytopenia

Congenital amegakaryocytic thrombocytopenia is caused by the inheritance of a mutation in the MPL gene, which is involved in the production of platelets. This affects the coagulation mechanism. Affected children will usually bruise easily, which prompts diagnosis early in life. It can eventually affect the production of all blood cells and may increase the risk of leukemia.

Severe Chronic Neutropenia

Severe chronic neutropenia affects the white blood cells, in particular the neutrophils. It is usually diagnosed in early childhood at a mean age of three years. It is caused by one of several inherited gene mutations that affect white blood cell production. Affected children are prone to infections, mouth ulcers and gum disease.

Thrombocytopenia Absent Radii Syndrome

The thrombocytopenia absent radii syndrome is characterized by the absence of the radius bone in each lower arm. Affected children usually bruise easily, due to the decreased production of platelets, which are crucial components in blood clotting. It is caused by a hereditary abnormality of the RBM8A gene, and is usually diagnosed in infants before the age of six months.

Acquired Aplastic Anemia

Aplastic anemia occurs due to an acquired rather than inherited gene mutation, and is usually diagnosed in adults, although it can sometimes also affect children. The mutation is usually acquired following exposure to radiation, chemicals or infections. It severely impairs the production of all blood cells and can lead to symptoms such as fatigue, dizziness, bleeding and increased susceptibility to infections. Paroxysmal nocturnal hemoglobinuria (PNH) is a type of acquired aplastic anemia that affects the bone marrow and blood stem cells.

Reviewed by Liji Thomas, MD.

References

- https://www.mskcc.org/pediatrics/cancer-care/types/pediatric-blood-disorders/about-pediatric-blood-disorders
- http://www.danafarberbostonchildrens.org/conditions/blood-disorders.aspx
- http://www.chw.org/medical-care/macc-fund-center/conditions/hematology-and-blood-disorders/blood-disorders/anemia-in-children/
- https://www.mdanderson.org/cancer-types/childhood-hematology-disorders.html

Further Reading

- What is Hematology?
- Hematology Tests
- Hematology Treatments

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