HEMOGLOBINOPATHY

[1]Sultonova Mukhlisa, [2]Sultonova Maftuna, [3]Islomjonova Iroda, [4]Mirzakeldiyeva Khatira, [5]Naimov Bunyod

[1,2,3,4]The students of Tashkent Pediatric Medical Institute

[5]Research advisor, The assistant teacher of Tashkent Pediatric Medical Institute, The Department: Medical and biological chemistry, medical biology, general genetics

https://doi.org/10.5281/zenodo.11097743

Abstract*. Hemoglobinopathy is a group of hereditary blood diseases and pathologies that primarily affect red blood cells. These are monogenic disorders and in most cases they are inherited as autosomal dominant traits*

Keywords*: hematology; hemoglobin; molecular diagnosis; sickle cell disease; thalassemia; β-thalassemia intermedia.*

INTRODUCTION

There are two main groups of hemoglobinopathies: abnormal structural variants of hemoglobin, caused by mutations in the genes encoding it, and thalassemias, which are caused by insufficient synthesis of normal hemoglobin molecules. The main structural varieties of hemoglobin are HbS, HbE and HbC. The main types of thalassemia include alpha thalassemia and beta thalassemia.

These two pathologies can be combined because some conditions that cause abnormalities in the structures of hemoglobin proteins also affect their synthesis. Some structural variants of hemoglobin do not cause pathology or anemia and therefore are often not classified as hemoglobinopathies.

MATERIALS AND METHODS

Normal variants of human hemoglobins are tetrameric proteins that contain two pairs of globin chains, each consisting of one alpha-like (α-like) chain and one beta-like (β-like) chain. Each globin protein chain is associated with an iron-containing heme moiety. Throughout life, the synthesis of alpha-like and beta-like (also called non-alpha-like) chains is balanced, so that their ratio is relatively constant and there is no excess of either type

The specific α- and β-like chains that are included in hemoglobin are highly regulated during development:

Fetal hemoglobins are expressed as early as the fourth to sixth week of embryogenesis and disappear around the eighth week of pregnancy as they are replaced by fetal hemoglobins. Fetal hemoglobins include:

Hb Goover-1, consisting of two ζ-globins (zeta-globins) and two ε-globins (epsilon globins) (ζ2ε2)

Hb Goover-2, consisting of two alpha globins and two epsilon globins (α2ε2)

Hb Portland, consisting of two zeta globins and two gamma globins (ζ2γ2)

Thalassemias are quantitative defects that result in decreased levels of one type of globin chain, creating an imbalance in the ratio of alpha-like to beta-like chains. As noted above, this ratio is normally tightly regulated to prevent the accumulation of excess globin chains of one type. Excess chains that are not incorporated into hemoglobin form nonfunctional complexes that are

deposited in erythrocytes. This can lead to premature destruction of red blood cells in the bone marrow (beta thalassemia) and/or in the peripheral blood (alpha thalassemia).

RESULT AND DISCUSSIONS

Transport of oxygen from the lungs to the tissues: This is due to the special interaction of the globin chains, which allows the molecule to absorb more oxygen where the oxygen content is high and release oxygen when the oxygen concentration is low.

Transfer of carbon dioxide from tissues to lungs: The end product of tissue metabolism is acidic, which increases the content of hydrogen ions in solution. Hydrogen ions combine with bicarbonates to form water and carbon dioxide. Carbon dioxide is absorbed into hemoglobin, facilitating this reversible reaction.

Nitric oxide transport: Nitric oxide is a vasodilator compound. This helps in regulating the vascular response during times of stress, such as inflammation.

Various structural abnormalities can lead to any of the following pathological processes:

Anemia due to shortened red blood cell lifespan or decreased production of cells such as hemoglobin S, C and E.

Increased affinity for oxygen: Red blood cells do not readily release oxygen under hypoxic conditions. Consequently, the bone marrow must produce more red blood cells, which leads to the development of polycythemia.

Unstable hemoglobin: Red blood cells are easily destroyed under stress and hemolysis occurs with the possible development of jaundice.

Methemoglobinemia: The iron in the heme portion of hemoglobin is easily oxidized, reducing the hemoglobin's ability to bind oxygen. More deoxygenated hemoglobin is produced and the blood becomes cyanotic.

CONCLUSION

Why we need to learn it -Some hemoglobinopathies (and related diseases such as glucose-6-phosphate dehydrogenase deficiency) appear to have conferred an evolutionary advantage, especially to heterozygous organisms, in areas where malaria is endemic. Malarial plasmodia live inside red blood cells but interfere with their function. In patients predisposed to rapid red blood cell clearance, this may lead to early destruction of cells infected by the parasite and increase the chances of survival of the carrier of this trait.

REFERENCES

1. CDC. Hemoglobinopathies Research. Centers for Disease Control and Prevention (February 8, 2019). Retrieved May 5, 2019. Archived May 5, 2019.
2. Weatherall D. J., Clegg J. B. Inherited haemoglobin disorders: an increasing global health problem. (English) // Bulletin Of The World Health Organization. - 2001. - Vol. 79, no. 8. - P. 704-712. - PMID 11545326.
3. Hemoglobinopathies and Thalassemia. medicalassistantonlineprograms.org/. Retrieved September 30, 2022. Archived from the original on January 9, 2015.
4. Hemoglobin Variants. Lab Tests Online. American Association for Clinical Chemistry (November 10, 2007). Access date: October 12, 2008. Archived November 5, 2006.
5. Huisman THJ. A Syllabus of Human Hemoglobin Variants. Globin Gene Server. Pennsylvania State University (1996). Access date: October 12, 2008. Archived September 1, 2006.