HERMANSKY – PUDLAK SYNDROME: REPORT OF A CASE AND REVIEW OF THE LITERATURE

Ilhan G1*, Acipayam C2, Kaya H3

1. Hematology Unit, Hatay Antakya State Hospital, Turkey
2. Pediatric Hematology, Mustafa Kemal University, Turkey
3. Hematology Unit, Mustafa Kemal University, Turkey

Correspondence: Dr Gul Ilhan. Hematology Unit, Hatay Antakya State Hospital, Turkey Email: gullhan2002@yahoo.com


ABSTRACT

Hermansky-Pudlak Syndrome (HPS) is rare, autosomal recessive disorder which is characterised with oculocutaneous albinism, abnormal platelet function and lysosomal ceroid lipofuscin accumulation. We presented a HPS case with bleeding attacks after breast reduction operation.

Key words: Hermansky-Pudlak Syndrome, platelet, function

INTRODUCTION

Hermansky-Pudlak syndrome is rare, autosomal recessive disease and described by two Czechoslovakian pathologists named Hermansky and Pudlak in 1959. Oculocutaneous albinism and prolonged bleeding time are the characteristic features of the disease. Lysosom related organels such as platelet dense granuls and melonosome are synthesesd abnormally. There are 8 subtypes of the disease. Platelet aggregation defect, congenital nistagmus, oculocutaneous albinism, iris transullumination, hypopigmented skin and hair are seen in this disease. Pulmonary fibrosis, granulamatous colitis and neutropenia are seen in some subtypes. These subtypes are distinguished by molecular analysis1–2. There are no curative treatment of the disease. Supportive therapy can be made. We reported a HPS case presenting with recurrent bleeding in surgical area.

CASE REPORT

A 30 year old women was admitted to hospital for breast reduction operation. She has no bleeding diathesis history except for prolonged menstruel bleeding. Preoperative tests including platelet number and prothrombin time were normal. During operation, surgeons had difficulty for stopping her bleeding. She had many units of erytrocytes and fresh frozen plasma. She was reoperated because of bleeding of the operation area in postoperative 30th day. She had two more revision operation similarly. She was
consultated with Hematology. Oculocutaneous albinism and nistagmus were found after physical examination. Her sister and brother had similar findings. Her platelets were 200,000/µL, APTT: 32 , PTZ: 11, INR: 1.1. In platelet aggregation tests, primary aggregation with collagen and epinephrine, ristosetin was normal, secondary aggregation with arachidonic acit was abnormal. We diagnosed her HPS. In Hematology policlinic control, we stopped operation area bleeding with ankaferd blood stopper.

DISCUSSION

Oculocutaneous albinism is the most important finding in this disease. Female patients are generally presented with prolonged menstruel bleedings. These patients have horizontal nystagmus as other albinism types. Complications of HPS are granulamatous colitis, renal dysfunction, pulmonary fibrosis and neutropenia. HPS is autosomal recessive disease and has 8 forms. HPS-1 is the most frequent and severe form. HPS - 4 is also clinically severe type. Other forms of the disease are seen as less severe and less frequent. HPS is one of the subtypes of platelet depot diseases which are seen with deficiency of alpha and dense granules. ADP, ATP, calcium and serotonin play role in secondary aggregation and they are in dense granules. In HPS, primer aggregation with ADP and epinephrin is normal but secondary aggregation tests are abnormal. HPS patients are generally diagnosed in early childhood with bleedings of soft tissues, nose as well as during surgery and childbirth. Coagulation factors levels, prothrombin time and partial thromboplastin time are normal. Dense granules deficiency can be seen by electron mycrospose. In HPS, there is dysfunction of lysosome related aganells such as melonosome, platelet dense granules and lameller bodies of type II alveol cells. Dysfunction of melonosomes causes oculocuaneous albinism and visual disorders. Pulmonary fibrosis mechanism can not be understood. Ceroid lipofuscin is may be the underlying cause. This pigment accumulates along life normally. But in some HPS patients, there is excessive accumulation. This accumulation damages type II alveolar cells and chronic inflammation, pulmonary fibrosis occur. This deposition causes renal dysfunction and granulomatous colitis in some patients. In addition, there may be moderate accumulation in bone marrow, spleen, liver, colon and other tissues. In practice, it is not necessary to show lipit-protein complex for diagnosis. There is no definite treatment of HPS. Mortality from bleeding of the disease is not high. Anemia can be seen after bleedings. Thrombocyte or whole blood transfusion may be done during bleedings with teeth extraction, birth or surg cal prosedures. Thrombocyte or whole blood transfusion can be done during bleedings of teeth extraction, birth or surgical prosedures. Desmopressin can be given for prophylaxis. There are several reports showing recombinant factor VII (VIIa) stopped bleeding. Aspirin using should be avoided.

CONSENT

Written informed consent was obtained from the patient for publication of this case report.
COMPETING INTERESTS

The authors declare that they have no competing interests.

REFERENCES