Paroxysmal nocturnal hemoglobinuria (PNH) and G6DP deficiency (two hemolytic anemias) on same patient

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Abstract

Background: Paroxysmal Nocturnal Hemoglobinuria (PNH) is a rare acquired hematopoietic stem cell disorder. They also have a tendency for thrombosis with predilection for unusual locations in the venous system. We report a young Filipino lady who is having two types of hemolytic anemias PNH and G6DP. We find this according to our knowledge as extremely rare.

Case scenario: 31 Year Filipino ladies admitted in 2016 with the chief complaints of red colored urine and yellowish discoloration of eyes; she also had episodes of dizziness, generalized weakness, headache and SOB. Investigation on this admission showed anemia, peripheral smear revealed a picture of hemolysis again. Direct Antiglobulin Test (DAT) was ordered and it came out as negative and subsequently the patient received blood transfusions. She had hemolysis as evidenced by raised LDH, low haptoglobin and raised bilirubin in the absence of a positive DAT test; this made us to suspect PNH. In order to confirm the same, we did flow cytometry which showed large (PNH) clone within the red cells (69.6%), granulocytes (99%) and the monocytes (98%). The hemolysis work up at the same time showed low G6DP. Eculizumab requested for patient but meanwhile they treated with steroids and her hemoglobin improved and hemolysis stopped.

Discussions PNH is classified into classic PNH (presence of hemolysis with no marrow abnormality); PNH with marrow disorders (aplastic anemia/myelodysplastic syndrome /primary myelofibrosis and subclinical PNH without clinical evidence. G6PD is needed to maintain NADPH and consequently reduced glutathione levels in red blood cells. G6PD-deficient people, mainly males can be asymptomatic but they are subjected to episodes of hemolysis, when the red blood cells are subjected to oxidative stress caused by infections, certain drugs or in the case of favism, after eating fava beans. Several polymorphic variants have been described with specific geographical distributions, though milder than other variants such as G6PD. G6PD deficiency usually only affects hemizygous males and homozygous females but heterozygous females can be affected when for example; biased X-inactivation has led to a predominance of red blood cells expressing the mutant protein. In PNH there is complement induced lysis of RBCs due to the abnormal sensitivity of RBC cell membrane. Hemolysis this causes reduction in hemoglobin and hemoglobinuria with resultant increase in LDHPNH is X linked and so do G6DP, so we suggest and hypothesis that chromosome x has role in this rare case of combined hemolytic anemia. Another interesting thing to observe or mention according to our literature review was all previous two cases in addition to our case were females.

Conclusion: Association of G6DP and PNH is very rare. There is no clear evidence so far, but the hypothesis it related to chromosome X and further case reports and studies are needed to study in this association.

Recent Publications

1. Shehab F Mohamed, Mohamed A Yassin, Abdulqadir J Nashwan and Susana E L akiki (2018) Concomitant BRAF mutation in hairy cell

leukemia and papillary thyroid cancer. Blood DOI: 10.1182/blood-2018-99-116084.

2. Shehab Fareed, Abdulqadir J Nashwan, Sulieman Abu Jari and Moh Yassin (2017) Spinal Abscess Caused by Salmonella Bacteremia in a Patient with Primary Myelofibrosis. American Journal of Case Reports 18:859-864.

Biography

Shehab F Mohamed is a Hematologist in National Center for Cancer Care and Research, Qatar which is a part of Hamad Medical Corporation. He has multiple publications in infections disease, hematology leukemia, MPNs.

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Notes/Comments: