ORIGINAL RESEARCH PAPER

Paediatric Medicine

G6PD DEFICIENCY IN A FEMALE CHILD:A RARE CASE REPORT

KEY WORDS:

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INTRODUCTION

G6PD deficiency is a genetic disorder that affects the enzyme glucose -6 -phosphate dehydrogenase leading to hemolytic anemia. It is the mostcommon enzymatic deficiency worldwide and accounts for 400 million people around the world,whichcontributes10%of world population. It has two variants–mediterranean variant(mild form)and African variant(severe form). It is also known as favism. It is an x -linked recessive disorder ,males are commonly affected, rare in females-except x chromosome inactivation (lyonization). females can be normal, deficient (homozygous)or intermediate(heterozygous).

Biochemical Aspect

1. It protects the cells from free radicals (e.g. H_2O_2).
2. It protects the cells from oxidative agents or stressor (Eg. Sulpha drugs: they can produce free radicals which can convert normal hemoglobin to met hemoglobin by oxidizing ferrous into ferric ion.NADPH will convert Met hemoglobin to normal Hb ,hence protects our cells from free radicals
3. Oxygen dependent killing by WBC(NADPH can convert O_2 to superoxide, which could help WBC to kill bacteria).

Case Presentation

A 7month old well thrived female patient was already diagnosed with G6PD deficiency during neonatal jaundice evaluation at neonatal period and presented with complaints of 2 episodes of GTCS convulsion associated with fever. General and systemic examinations were in normal range for age, no organomegaly. G6PD enzyme analysis was repeated and found out to be G6PD deficient. Patient was treated for meningitis after CSF analysis. karyotyping was done and found out to to be normal female karyotype, hence Lyon's hypothesis has been ruled out. Both parents were also G6PD deficient on subsequent enzyme analysis. Patient got both deficient x genes from parents hence without Lyon's hypothesis, XX female got G6PD deficiency.

Pathophysiology

Oxidative agents causes damage to RBC ,as they lack G6PD enzyme and NADPH leading to intravascular hemolysis. Macrophages engulf these damaged RBCs. Hemoglobin is released, which binds to a plasma protein known as haptoglobin. Hemoglobin is disintegrate into heme and globin. Iron and protoporphyrin are released from heme.Iron is converted into hemosiderin and eliminate through kidney as hemosiderinuria. hemoglobin can also be excreted through kidney as hemoglobinuria. In liver protoporphyrin is ultimately converted into conjugated bilirubin and hence excreated.

Clinical Presentation & Diagnosis

Most patients are asymptomatic. It can be presents as acute hemolytic episodes or chronic non spherocytic hemolytic anemia. Symptoms can be related to anemia, jaundice or in the neonatal period presents as neonatal jaundice. Definitive diagnosis can be made by DNA testing and enzyme analysis. Hb level is decreased and there will be elevated levels of S.LDH and unconjugated bilirubin. Peripheral smear shows bizarre poikilocytosis, hemighost, Heinz bodies and bite cells. Urine examination reveals hemoglobinuria and hemosiderinuria.

CONCLUSION

It is the most common enzymatic deficiency and can be prevented by avoiding oxidative stressors. Screen for G6PD enzyme before starting dapsone, chloroquine, sulfasalazine, nitrofurantoin, ciprofloxacin, aspirin, Rasburicase. Blood transfusion in acute hemolytic episodes.

REFERENCES

[1] Nelson text book of pediatrics-21st and 22nd edition