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NEONATAL SCREENING FOR GLUCOSE 6 PHOSPHATE DEHYDROGENASE DEFICIENCY

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ABSTRACT

Background: Glucose 6 phosphate dehydrogenase(G6PD) deficiency , most common enzyme deficiency involving more than 400 million people worldwide causes a spectrum of disease including neonatal hyperbilirubinemia, acute hemolysis, and chronic hemolysis. **Aim:** To evaluate the incidence of G6PD deficiency in neonates in a tertiary care hospital. The incidence of hyperbilirubinemia in G6PD deficient subject was compared with that of hyperbilirubinemia in G6PD normal subject. **Materials and Methods :** In this prospective study all male and female neonates born during a period of one year in a tertiary care hospital were included. Qualitative study was done from blood samples taken from cord blood for screening of neonates to determine the incidence of G6PD deficiency . **Results :** Incidence of G6PD deficiency was found to be 1.92%. Incidence in male was 2.1% and incidence in female was 1.6%.Incidence of hyperbilirubinemia was found to be 41.6% in case of G6PD deficient subject which was much lower than the incidence of hyperbilirubinemia in G6PD normal subject which was found to be 15.3% . **Conclusion :** We conclude that neonatal screening for G6PD deficiency is an useful test for prevention and early treatment of complications which are associated with it.

KEYWORDS Glucose 6 Phosphate dehydrogenase deficiency,hyperbilirubinemia,neonatal screening.

INTRODUCTION

G6PD is a house keeping enzyme which catalyzes the first step in the pentose phosphate pathway . The G6PD gene is located on the telomeric region of the long arm of Xchromosome (Xq28) and is 18 kb long consisting of 13 exons, transcribed to a 2.269 kb mRNA with 1.54 kb of coding regions .The main physiological role of G6PD is to provide NADPH, a compound necessary for a number of detoxification and biosynthetic reactions, including fatty acid synthesis. Thus, lack of G6PD enzyme in the red blood cells is lethal and deficiency in the enzyme in case of oxidative stress is deleterious to the cell.[1] G6PD deficiency is the commonest enzyme disorder of human beings and a globally important cause of neonatal jaundice, which can lead to kernicterus and death or spastic cerebral palsy. It can also lead to life-threatening haemolytic crises in childhood and at later ages, by interacting with specific drugs and with fava beans in the diet.[2] This X-linked inherited disorder most commonly affects persons of African, Asian, Mediterranean, or Middle-Eastern descent. Approximately 400 million people are affected worldwide. Homozygotes and heterozygotes can be symptomatic, although the disease typically is more severe in persons who are homozygous for the deficiency.[3] The complications of G6PD deficiency can largely be prevented by education and information, and neonatal jaundice can be successfully treated by phototherapy, a cheap and simple approach suitable for use in primary health care.[2] **Aims**: This study was done to evaluate the incidence of glucose 6 phosphate dehydrogenase deficiency in neonates in a tertiary care hospital and to compare the the incidence of hyperbilirubinemia in G6PD deficient subject with the incidence in G6PD normal subjects. **Materials and Methods** : In this prospective study all male and female neonates born during a period of one year in a tertiary care hospital were included. Qualitative study (Kit-Autozyme new G6PDH Qualitative dye method by Accurex) was done from blood samples taken from cord blood for screening of neonates to determine the incidence of glucose 6 phosphate dehydrogenase deficiency. Whole blood was collected in a clean dry container using EDTA as an anticoagulant. Heparin should not be used as it interferes with the reaction. For an unknown sample the haemoglobin content must first be estimated and aliquot of blood may be corrected for low haemoglobin content. Haemoglobin content of whole blood was estimated. If the haemoglobin content is significantly less than 15 gm/dl, the haemoglobin content is adjusted by proportionately increasing the aliquot of whole blood during preparation of red cell haemolysate.

Principle of the test-Glucose 6 phosphate dehydrogenase (G6PD) present in haemolysate acts on substrates,Glucose-6-phosphate and NADP, giving NADPH, which in presence of Phenazine methosulfate decolorises blue coloured indophenol dye leaving behind colour only due to haemolysate. The rate of reaction is proportional to G6PDH present ,hence time required for decolorisation is inversely proportional to G6PDH activity in the haemolysate.

Glucose-6-Phosphate+NADP----------◊6-Phosphogluconate+ NADPH

DCPIP+NADPH----------◊Reduced DCPIP

Reaction mixture is observed at 30 minutes for decolourisation. If the decolourisation is complete, it is observed every 5 minutes there after until the decolourisation is complete. In G6PDH deficiency the time taken for decolourisation will exceed from 2 hours to 24 hours.

In normal subjects, decolourisation time is between 30-60 minutes.

In G6PDH deficient subjects, (heterozygous males and homozygous females) decolourisation time is between 2-24 hours.

In heterozygous females, who are carriers, the cell population is mixed with normal and deficient cells. The distribution of deficient cells varies from 20% to 80%.Hence some subjects may give results overlapping over normal as well as abnormal time specifications.

Bilirubin level in all the neonates was measured. Other factors causing causing neonatal hyperbilirubinemia such as sepsis, ABO and Rh incompatibility, infant of diabetic mother, polycythemia were excluded from the study. Results: A total of 1250 neonates were included in this study. There were 750 males and 500 females.24 neonates were found to be G6PD deficient. Incidence of G6PD deficiency was found to be 1.92%. 16 males and 8 females were found to be G6PD deficient ,incidence being 2.1% in

males and 1.6% in females. Thus the incidence of G6PD deficiency was higher in males than in females which was in accordance with other study. Out of 24 G6PD deficient neonates in this study,10 had hyperbilirubinemia, incidence being 41.6% while the incidence of hyperbilirubinemia in G6PD normal subjects was found to be 15.3% .Thus the incidence of hyperbilirubinemia in G6PD deficient subjects was higher than in G6PD normal subjects.

Conclusion: We conclude that neonatal screening for G6PD should be done routinely so that we can manage the complications at the earliest.

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