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STUDY ON SCREENING FOR GLUCOSE 6 PHOSPHATE DEHYDROGENSASE DEFICIENCY IN NEWBORN BABIES AT OUR TERTIARY CARE HOSPITAL

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ABSTRACT

Background: Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common human enzyme deficiency in the world. G6PD deficiency is an X-linked disorder affecting mostly African, south east Asian and middle-eastern population. In India, there are 13 biochemical variants of G6PD being reported so far out of which Mediterranean type is most common in the caste groups. Aim and Objectives: to find out the incidence of G6PD deficiency in neonates delivered at our tertiary care hospital and to assess the contribution of G6PD deficiency in causing Neonatal jaundice. Materials and Methods: The study was a prospective hospital based type undertaken among the 400 babies delivered at our hospital from. The objective of this study was to find out incidence of G6PD deficiency in babies delivered in our Hospital and to assess its contribution in causing neonatal jaundice. A detailed antenatal history, consanguinity, geographical area of mother, birth order, mode of delivery, gestational age, birth injuries and blood group were recorded in each baby by interviewing the mother after taking consent in prescribed proforma. The following laboratory investigations were carried out, G6PD screening test (fluorescent spot test), Baby blood group and Rh typing, Mother blood group and Rh type and If jaundice present total, direct and indirect bilirubin testing was done. If the sample (cord blood) shows G6PD deficiency by fluorescent spot method, quantitative assay was done. Discussion and Conclusion: This study was conducted to detect incidence of G6PD deficiency in neonates and as a contributory factor in neonatal hyperbilirubinemia. The result of this study shows that incidence of G6PD deficiency in neonates delivered at our hospital is 3%. It contributes significantly for Neonatal Jaundice.

Key-words: Glucose-6-phosphate dehydrogenase (G6PD) deficiency, newborn babies, fluorescent spot assay and neonatal jaundice.

<u>INTRODUCTION</u>: Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common human enzyme deficiency in the world [1]. G6PD deficiency is an X-linked disorder affecting mostly African, south east Asian and middle-eastern population [2,3]. In India, there

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are 13 biochemical variants of G6PD being reported so far out of which Mediterranean type is most common in the caste groups. Orissa variant is most prevalent in tribals of India. Kerala-Kalyan is the 3rd common variant [4]. Incidence of G6PD deficiency in different geographical areas of India varies from 0 to 27% [5]. G6PD is the first enzyme of the pentose phosphate pathway and catalyzes the conversion of glucose-6-phosphate to 6-phosphogluconolactone (G6P), with the concomitant reduction of nicotinamide adenine dinucleotide phosphate (NADP) to its reduced form (NADPH). NADPH protects cells from oxidative stress. Glucose-6-phosphate dehydrogenase deficiency causes a spectrum of diseases including neonatal hyperbilirubinemia, acute hemolysis. This deficiency can cause hemolytic anemia, usually after certain medications. Homozygotes and heterozygotes can be symptomatic, although the disease typically is more severe in persons who are homozygous for the deficiency. The conversion of nicotinamide adenine dinucleotide phosphate to its reduced form in erythrocytes is the basis of diagnostic testing for the G6PD deficiency. This usually done by fluorescent spot test. With the right precautions, a child with G6PD deficiency can lead a healthy and active life [6]. Hence this study was done to know the incidence of G6PD deficiency in neonates delivered at our tertiary care hospital and its contribution in causing neonatal jaundice.

<u>AIM AND OBJECTIVES</u>: the objectives of the study were to determine the incidence of G6PD deficiency in neonates delivered at our tertiary care hospital and to assess the contribution of G6PD deficiency in causing Neonatal jaundice.

MATERIALS AND METHODS:

The study was a prospective hospital based type undertaken among the 400 babies delivered at our hospital from September 2021 to August 2022. The objective of this study was to find out incidence of G6PD deficiency in babies delivered in our Hospital and to assess its contribution in causing neonatal jaundice.

A detailed antenatal history, consanguinity, geographical area of mother, birth order, mode of delivery, gestational age, birth injuries and blood group were recorded in each baby by interviewing the mother after taking consent in prescribed proforma.

Study design: Prospective hospital based study.

Sample size: 400 babies delivered in our hospital in the duration of one year.

Inclusion Criteria: Babies delivered at our hospital.

Exclusion Criteria: none

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Blood Sample Collection and Biochemical Investigations: The following laboratory investigations were carried out,

- 1. G6PD screening test (fluorescent spot test)
- 2. Baby blood group and Rh type
- 3. Mother blood group and Rh type
- 4. If jaundice present
- a. Indirect bilirubin
- b. Direct bilirubin
- c. Hemoglobin
- d. DCT (direct Coomb's test)
- e. Retic count

If the sample (cord blood) shows G6PD deficiency by fluorescent spot method, quantitative assay was done. 1ml cord blood of all babies collected in EDTA vials immediately after birth. 10 microliters of collected blood is added to 100 microliter reagent mixture, a spot is made on a Whatman No.3 filter time. The sample is incubated at room temperature and further spots made at 5,10,15, and 20 minutes respectively. The spots are allowed to dry and examined under a long wave UV lamp (Fluorescent Spot Test Method). In normal samples, the first spot fluoresces slightly with increasing in fluorescence in the remaining spots indicating reduction of NADP to NADPH+H. G6PD deficient sample did not show fluorescence in any of the spots. Babies found to have G6PD deficiency were followed, if they develop jaundice standard management protocol of the hospital is employed.

Criteria for some of the variables are: Cord blood samples fluoresce under fluorescent microscopy were considered as normal G6PD activity. Preterm babies had been defined as those neonates having gestational age < 37 weeks, and low birth weight (LBW) babies were those whose birth weight <2.49 kg. Serum bilirubin levels were obtained when newborns developed jaundice.

Descriptive statistics:

The descriptive procedure displays univariate summary statistics for several variables. In a single table and calculates standardized values (Z score). Variables were ordered by the size of their means (in ascending or descending order), alphabetically, or by the order in which we select the variable (the default)

Cross tabs procedure:

The crosstabs procedure forms two-way and multiway tables and provide a variety of tests and

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measure of association for two-way tables. The structures of the table and whether categories are ordered determine what test or measure to use. All the statistical methods were carried out through the SPSS for windows.

<u>RESULTS</u>: In the present study, 400 babies delivered in our hospital, were included for screening for G6PD deficiency by fluorescent spot method, we found that 12 babies had screening positive and the incidence of G6PD was 3%.

Table 1: Incidence of G6PD deficiency in neonates delivered in our hospital					
Number of babies	Method	Number of babies with G6PD deficiency	Incidence		
400	Fluorescent spot method	12	3%		

Table 2: Sex of the baby versus G6PD screening					
Sex of the baby	Frequency	Percentage	G6PD (normal)	G6PD (deficient)	
Male	232	58	225	11	
Female	168	42	163	1	
Total	400	100	388	12	

Table 3: Causes of Jaundice Versus G6PD Deficiency			
Cause of Jaundice	Number of Babies		
Rh Incompatibility	6		
OA Incompatibility	6		
Infants of Diabetic Mother	2		
Sepsis	2		
Exaggerated physiological jaundice	22		
G6PD Deficiency	12		
Total	50		

DISCUSSION:

In the present study, 400 babies delivered at our hospital were screened to detect incidence of G6PD deficiency in neonates by fluorescent spot assay. Out of the 400 babies screened 12 babies had G6PDH deficiency by fluorescent spot assay, the incidence is found to be 3%.

In study done by Mritunjay et al, [7] out of 2479 babies screened for G6PD deficiency by a semiquanatitive assay, 50 neonates were found to be G6PD deficiency with incidence being 2%. Ramin Iranpour et al [8] screened 2501 babies, by a semiquanitative assay, out of which 79 neonates were found to have G6PD deficiency, incidence being 3.2%.

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In the present study, out of 400 babies, 232 babies (58%) were males and 168 were females (42%), in the present study we found the Incidence of 3%. In study done by Mritunjay et al [7] out of 2479 babies screened for G6PD deficiency, 1343 were males and 1136 were females. 2.83% males and 1.05% females were found to be G6PD deficient. Ramin Iranpour et al [9] screened 2501 babies, 1307 were males and 1194 were females. Out of which 67 males (5.1%) and 12 (1%) females were found to be G6PD deficient.

Regional distribution of G6PD deficiency studied by Bhasin et al, [10] showed 0% deficient in Karnataka, 0.03% in Andhra pradesh, and 0.07% in Tamilnadu. Study by Vandana Rai et al, [11] showed the frequency of G6PD deficiency among Indian population as a whole ranges from 0 to 27%. G6PD deficient frequency is comparatively higher in North and West Indian zones, whereas in South India it is uniformly low except in Andhra Pradesh and Tamil Nadu. Hakim et al screened 186 babies in Kerala, all were showing normal G6PD activity.

Various aetiologies of jaundice revealed that out of 400 babies 6 babies had Rh incompatibility, 6 had OA incompatibility, 2 babies were infants of diabetic mother, 2 babies had sepsis, 22 had exaggerated physiological jaundice, 12 babies had G6PDH deficiency.

CONCLUSION:

In the present study, 400 babies delivered in our hospital were screened to detect incidence of G6PD deficiency in neonates. The screening was done by fluorescent spot test. The incidence of G6PD deficiency found to be 3% among 400 babies screened.

This study was conducted to detect incidence of G6PD deficiency in neonates and as a contributory factor in neonatal hyperbilirubinemia. The result of this study shows that incidence of G6PD deficiency in neonates delivered at our hospital is 3%. G6PD deficiency contributes as one of the significant cause for neonatal jaundice in our hospital.

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