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A STUDY OF CLINICAL PROFILE OF GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY IN NEONATES PRESENTED WITH INDIRECT HYPERBILIRUBINEMIA

Paediatric Medicine

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ABSTRACT

Background: G6PD deficiency shows X-linked recessive inheritance [1]. It can cause a spectrum of diseases including neonatal jaundice, acute haemolysis, and chronic haemolysis [1]. Sudden episodes of haemolysis associated with this condition increase the serum bilirubin to high levels inducing neurologic damage [2,3]. **Materials And Methods:** The observational descriptive study aimed to determine the prevalence of G6PD deficiency in neonates presented with indirect hyperbilirubinemia, their clinical profile and comparison of clinical profile with non-deficient neonates. The study was undertaken 210 neonates with indirect hyperbilirubinemia in SNCU of Bankura Sammilani Medical College. **Result:** The prevalence of G6PD deficiency in neonates who presented with indirect hyperbilirubinemia is 15.23%. There was statistical significance found in between G6PD deficient and non-G6PD deficient groups in terms of male gender, higher total and direct serum bilirubin levels, lower haemoglobin levels and longer duration of phototherapy requirement. However, there was no statistical difference in terms of early age of onset of jaundice, religion, total leucocyte count, platelet count, ESR, reticulocyte count and requirement of PRBC transfusion and exchange transfusion. **Conclusion:** Early neonatal screening programs should be instituted to anticipate and start early treatment to prevent neonatal morbidity and mortality where G6PD deficiency prevalent among general populations.

KEYWORDS

G6PD (Glucose-6-Phosphate Dehydrogenase) Deficiency, Indirect Hyperbilirubinemia, Neonatal Jaundice, Hemolysis.

INTRODUCTION:

Neonatal indirect hyperbilirubinemia is one of the most common causes of morbidity in the first few days of life. This condition affects 60% of term and 80% of preterm babies [4,5]. Neonatal hyperbilirubinemia carries a substantial risk of harmful complications, including long-term neuronal complications due to kernicterus and even death [6]. G6PD enzyme is a part of Pentose Monophosphate Shunt. It catalyzes the oxidation of glucose-6-phosphate and the reduction of NADP+ to NADPH+. NADPH maintains glutathione [GSH] in its reduced form which helps in scavenging oxidative free radicals. RBCs do not generate NADPH in any other way so they are more susceptible to destruction from oxidative stress [7]. G6PD Deficiency shows X-linked recessive inheritance. Most babies with G6PD deficiency have a lack of symptoms. However, in symptomatic neonates, the presenting features are neonatal jaundice and/or acute haemolytic anaemia. Jaundice usually appears in 1-4 days, at the same time or slightly earlier than physiological jaundice. The marked elevation of bilirubin levels that sometimes occurs during neonatal period raises the risk of kernicterus in patients with G6PD deficiency[2,3].

AIM OF THE STUDY-

To study the prevalence of G6PD deficiency in neonates presented with indirect hyperbilirubinemia and to compare the course of neonatal jaundice in G6PD deficient versus non deficient neonates.

MATERIALS AND METHODS:

A total of 210 neonates with indirect hyperbilirubinemia were screened for G6PD deficiency from September 2022 to March 2024 in SNCU of Bankura Sammilani Medical College and Hospital, Bankura. A written consent was obtained before study.

Inclusion Criteria-

Both inborn and outborn babies presenting with indirect hyperbilirubinemia with serum bilirubin >5mg/dl were included in the study.

Exclusion Criteria-

- Neonates with serum bilirubin level ≤ 5mg/dl.
- Parents / guardians who did not give consent.
- Neonates presented with direct hyperbilirubinemia (direct bilirubin>1mg/dl if total bilirubin ≤ 5mg/dl or >20% if total bilirubin >5mg/dl)
- Neonates who received blood transfusion before determining G6PD status were excluded to avoid false results.

All cases were subjected to detail history and clinical examination as per predesigned pro forma. Each neonate was subjected to blood test which included-

- Complete Blood Count (CBC) with reticulocyte count.
- Total Serum Bilirubin and Direct & Indirect fraction of Bilirubin
- G6PD estimation.
- Blood grouping of mother and newborn.
- Direct coombs test.
- Reducing substance in urine.
- Thyroid profile (After Day 3 OF life).

Study Variables-

* Age at onset of Neonatal indirect hyperbilirubinemia. (Day of life)
* Sex distribution
* Religion
* Laboratory investigations variables-

a. TSB (total serum bilirubin) with indirect and direct proportion.
b. CBC (complete blood count)- Haemoglobin, TLC, Platelets with ESR and Retic count.
c. Blood for G6PD estimation ((G6PD activity below 7.24U /g of haemoglobin is considered significant; normal 8.83± 1.59U / g of haemoglobin).

*Treatment variables –
1. Duration of phototherapy
2. Need of Exchange transfusion.
3. Need of Blood Transfusion.

Statistical Analysis Plan

- Data was analyzed using appropriate statistical tests. The statistical analysis was carried out using available standard statistical software (SPSS 25 version).
- Quantitative variables were described by mean and standard deviation.
- The comparison of normally distributed continuous variables between the groups was done using Student's T test.
- Nominal categorical data between the groups was compared using Chi-squared test.
- P value <0.05 was considered significant

RESULTS-

The prevalence of G6PD deficiency in neonates who presented with indirect hyperbilirubinemia is 15.23%. (32 among 210 neonates), There was statistical significance found in between G6PD deficient and non-G6PD deficient groups in terms of male gender, higher total and direct serum bilirubin levels, lower haemoglobin levels and longer duration of phototherapy requirement. However, there was no statistical difference in terms of early age of onset of jaundice, religion, total leucocyte count, platelet count, ESR, reticulocyte count and requirement of blood transfusion (PRBC) and exchange transfusion.

DISCUSSION-

Among 210 patients in our study, 32 patients were G6PD deficient

neonates presented with indirect hyperbilirubinemia. The prevalence of G6PD deficiency in INDIA is from 2.3 to 27% with an average prevalence of 7.7% [8]. The prevalence of G6PD deficiency in neonates with indirect hyperbilirubinemia varies

Table 1: Clinical Profile And Comparison Between G6pd Deficient Vs Non-deficient Ones Presente With Inirect Hyperbilirubinemia (n=210)

VARIABLES	G6PD DEFICIENT (N1= 32)	G6PD NON-DEFICIENT(N2=178)	P Value
AGE (MEAN DAY±SD)	3.4±0.71	3.75±0.88	0.017(<0.05)
AGE(DAYS), n (%)			
<3 DAYS	06(18.8%)	12(6.7%)	0.069(>0.05)
3-5DAYS	25(78.1%)	154(86.6%)	
>5DAYS	01(3.1%)	12(6.7%)	
GENDER, n (%)			
MALE	31(96.8%)	94(52.8%)	< 0.00001
FEMALE	01(0.2%)	84(47.2%)	
RELIGION, n (%)			
HINDU	13(40.6%)	86(48.3%)	0.4(>0.05)
MUSLIM	19(59.4%)	92(51.7%)	
DURATION OF PHOTOTHERAPY(DSPT) MEAN±SD	4.15±1.09	3.43±0.98	0.001(<0.05)
DAYS, n(%)			
<3 days	4(12.5%)	38 (21.3%)	0.002(<0.05)
3-5 days	24(75%)	132 (74.2)	
>5days	4(12.5%)	08(4.5%)	
DVET EXCHANGE TRANSFUSION, n (%) REQUIRED	03(9.4%)	05(2.8%)	0.073(>0.05)
NOT REQUIRED	29(90.6%)	174(97.2%)	
PRBC TRANSFUSION, n (%) REQUIRED	03(9.4%)	05(2.8%)	0.073(>0.05)
NOT REQUIRED	29(90.6%)	174(97.2%)	

PRBC- PACKED RED BLOOD CELL, DVET- DOUBLE VOLUME EXCHANGE TRANSFUSION, DSPT- DOUBLE SURFACE PHOTOTHERAPY.

From the Table 1, it shows that although the mean age of onset of jaundice was significantly earlier in G6PD-deficient neonates (3.40 ± 0.71 vs 3.75 ± 0.88 days; p = 0.017), the categorical distribution of age of onset did not differ significantly between the two groups ($\chi^2 = 5.36$, p = 0.069). This finding should be underline to state the importance of early screening programs for neonatal morbidity and mortality.

Our study shows that all most all G6PD deficient neonates are males.

There was no statistical difference between G6PD deficient and non-deficient ones in terms of religion.

Although there was higher proportion of DVET an PRBC transfusion among G6PD deficient neonates but there was found no statistical difference between G6PD deficient and non-deficient neonates.

The duration of phototherapy was significantly longer in G6PD-deficient neonates compared to non-deficient neonates. Categorical analysis showed a significantly higher proportion of G6PD-deficient neonates requiring prolonged phototherapy (>5 days) ($\chi^2 = 12.6$, $p < 0.001$). Additionally, the mean duration of phototherapy was significantly greater in the G6PD-deficient group (4.15 ± 1.09 days vs 3.43 ± 0.98 days; $p < 0.001$).

In Table no.2 our study shows Mean total serum bilirubin was significantly higher in G6PD-deficient neonates compared to non-deficient neonates (22.29 ± 3.45 vs 19.45 ± 3.12 mg/dl; $p < 0.0001$). The distribution of total serum bilirubin categories also differed significantly between the two groups ($\chi^2 = 33$, $p < 0.0001$). Mean direct serum bilirubin was also showed significantly higher in G6PD-deficient neonates compared to non-deficient neonates (1.50 ± 0.52 vs 1.16 ± 0.55 mg/dl; p = 0.001). The distribution of direct serum bilirubin categories also showed a statistically significant difference between the two groups ($\chi^2 = 16$, p = 0.001). Mean haemoglobin level was significantly lower in G6PD-deficient neonates compared to non-deficient neonates (13.4 ± 2.55 vs 14.9 ± 2.27 g/dl; $p \approx 0.001$). The distribution of haemoglobin categories also differed significantly between the two groups ($\chi^2 \approx$ 13-14, p = 0.0009). This picture suggest that not only increase bilirubin secondary to haemolysis contribute to neonatal jaundice but also reduced hepatic conjugation and excretion of bile also contribute to same.

There was no significant association found between two groups in term of Total leucocyte count ($p \approx 0.06$) and Platelet count ($p \approx 0.64$).

Table 2: Comparsion Of Blood Parameters Between G6PD Deficient VS Non-deficient Ones Presente With Inirect Hyperbilirubinemia(n=210)

VARIABLES	G6PD DEFICIENT (N1= 32)	G6PD NON-DEFICIENT(N2=178)	P VALUE
TSB(MEAN±SD)	22.29 ±3.45	19.45±3.12	<.0001
TSB (mg/dl), n(%) ≤15	02(6.25%)	06(3.37%)	<.0001
15.1- 19.9	04(12.5%)	108(60.67%)	
20- 24.9	20(62.5%)	55(30.9%)	
≥25	06(18.75%)	09(5.06%)	
DSB(MEAN±SD)	1.50±0.52	1.16 ± 0.55	0.001(<0.05)
DSB (mg/dl), n(%) ≤1	06(18.76%)	97 (54.5%)	
1.1-1.4	11(34.37%)	55(30.9%)	
1.5-1.9	11(34.37%)	17(9.55%)	
≥2	04(12.5%)	09(5.05%)	
Hb(MEAN±SD)	13.4±2.55	14.9±2.27	0.001(<0.05)
Hb (g/dl), n(%) ≤ 10	03 (9.4%)	07(3.9%)	0.0009(<0.05)
10.1- 14.9	21(65.6%)	71(39.9%)	
≥15	08(25%)	100(56.2%)	
TLC (COUNT/µL) (MEAN±SD)	9.27K± 4.7K	7.9K±3K	0.06(>0.05)
PLATELET COUNT(LAKHS/µL) (MEAN±SD)	3.73±1.37	3.61± 1.30	0.64(>0.05)

ESR (mm/hr) (MEAN±SD)	8.56±3.37	7.48±3.30	0.09(>0.05)
RETICULOCTE COUNT (%) (MEAN±SD)	1.77±0.55	1.54±0.67	0.07(>0.05)

TSB- TOTAL SERUM BILIRUBIN, DSB-DIRECT SERUM BILIRUBIN, Hb-HAEMOGLOBIN, TLC- TOTAL LEUCOCYTE COUNT, ESR - ERYTHROCYTE SEDIMENTATION RATE G6PD- GLUCOSE-6-PHOSPHATE DEHYDROGENASE k- THOUSAND

Although mean ESR was slightly higher in G6PD-deficient neonates compared to non-deficient neonates (8.56 ± 3.37 vs 7.48 ± 3.30 mm/hr), the difference was not statistically significant ($p \approx 0.09$). Similarly, the distribution of ESR categories did not differ significantly between the two groups ($\chi^2 \approx 2.9$, $p = 0.091$).

Similarly, the mean reticulocyte count was higher in G6PD-deficient neonates compared to non-deficient neonates (1.77 ± 0.55 vs 1.54 ± 0.67%), the difference was not statistically significant ($p \approx 0.07$). Similarly, the distribution of reticulocyte count categories did not differ significantly between the two groups ($\chi^2 \approx 5.4$, $p = 0.0689$).

CONCLUSION-

G6PD deficiency is a common enzyme defect in neonatal populations, particularly in males, and causes severe indirect hyperbilirubinemia that requires treatment.

Neonatal jaundice with higher bilirubin assay and anaemia is more common in G6PD-deficient neonates.

Adequate and timely management should be provided to avoid irreversible neurological complications. Screening of newborns at risk is considered helpful, as early adequate treatment with phototherapy, exchange transfusion, and other medications can benefit their clinical outcome and prevent untoward complications.

LIMITATIONS-

Molecular studies to confirm the G6PD mutation were not performed in the G6PD deficient neonates due to the unavailability of such tests at our centre. The sample size enrolled for this study was limited due to time constraints, larger study required to validate the same.

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