ORIGINAL RESEARCH ARTICLE

INTERNATIONAL JOURNAL OF BIOASSAYS
ISSN: 2278-778X
CODEN: IJBNHY
OPEN ACCESS

Screening of G6PD deficiency in children presenting with anaemia and jaundice

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Received for publication: September 9, 2015; **Revised**: September 21, 2015; **Accepted**: October 5, 2015

Abstract: G6PD deficiency is one of the most common X-Linked hereditary disorder and enzymopathies in human that affect the erythrocyte metabolism with approximately 400 million people affected worldwide. G6PD deficiency most commonly manifests as either neonatal jaundice or acute haemolytic anaemia. About 400 different variants and ninety different mutations of this disease are known globally. The present study was conducted to screen children who are anaemic and jaundiced for this enzyme and to study the average haemoglobin and serum bilirubin levels in these children. Hundred children in various age groups were taken up. Their blood samples were analysed for G6PD enzyme, serum bilirubin and haemoglobin. Out of these 100 children, 52 were females and 48 were males. In our study 3 (3%) children (all males) were found to be G6PD enzyme deficient, age being 4 days, 3 days and 3 years. The commonest clinical presentation was jaundice followed by anaemia. Jaundice was found in 65 cases (65 %) with serum bilirubin levels above 1 mg/dl and anaemia was found in 55 cases (55 %). This study was undertaken in view of limited amount of data available for G6PD deficiency in children presenting with anaemia and jaundice in Jammu and surrounding regions, stressing on developing an early neonatal screening program to prevent significant morbidity and mortality in childhood.

Key Words: G6PD; Jaundice; Anaemia

INTRODUCTION

G6PD is the most common enzyme deficiency worldwide. It is an X-Linked Recessive disorder expressed mostly in males. G6PD deficiency causes a spectrum of diseases including neonatal hyper bilirubinemia, acute haemolysis and chronic haemolysis. According to the report of WHO[1], 7.5% of the world population carry one or two genes for G6PD deficiency and 2.9% are G6PD deficient. There are no primary prevention or interventions available for this disease and the only way to avoid the adverse outcomes is to recognize such children early in life and prevent exposure to agents which can trigger haemolysis[2]. Glucose 6 phosphate dehydrogenase is an enzyme of hexose mono phosphate (HMP) shunt which helps in maintaining the ionic millieu with in the RBC's and protects both haemoglobin and RBC membrane from exogenous oxidant stress, including certain drugs. When normal RBC is exposed to an offending drug, stress or toxin, the glucose metabolism via HMP shunt is increased several fold, which in turn generates reduced glutathione thus protecting sulfhydryl group of haemoglobin and RBC membrane from oxidation. G6PD deficiency is a group of hereditary abnormalities in which the activity of erythrocyte enzyme G6PD is markedly diminished. Such a deficiency may result in haemolytic anaemia, particularly after administration of drugs, during infections and in the neonatal period.

Individuals with an inherited defect in HMP shunt are unable to maintain an adequate level of reduced glutathione in their RBC's. As a result the Hb-sulfhydryl group becomes oxidised and the haemoglobin tends to precipitate with in RBC's forming heinz bodies. These RBC's become susceptible to haemolysis. About 400 different variants and 90 different mutations of this disease are known globally. The G6PD gene is located on the telomeric region of the long arm of X- chromosome (Xq28) and is 18 kb long consisting of 13 exons, transcribed to a 2.269kb mRNA with 1.545kb of coding regions. Inheritance pattern in G6PD deficiency is X- linked[3]. Therefore, theoretically speaking males suffer from the effects of deficiency and females are the carriers. G6PD deficiency occurs with increased frequency throughout Africa and Asia, the Mediterranean Middle East[4]. As India lies in a special geographic situation with different ethnic groups, the incidence of G6PD deficiency varies widely in different localities. Though the exact incidence in India is not known, various studies have reported an incidence ranging from 2-27.9% in different communities[5]. In India the most common mutation is the G6PD Mediterranean seen in the Vatalia Prajapatis of North India and the Parsis[6].

Common agents precipitating haemolysis in G6PD Deficiency are:

Drugs and Chemicals: Anti-Bacterials, Sulphonamides, Nalidixic acid, Chloramphenicol, Nitrofurantoin, Niridazole, Anti-Malarials**:** Primaquine, Chloroquine and Quinacrine

Others: Napthalene, Benzene, Methylene blue, Probenicid, Acetyl salicylic acid, Vitamin K analogues, Streptomycin, Sulfamethaxazole, Furazolidine

Illnesses: Infections, Diabetic Keto Acidosis, Hepatitis Certain Foods, Fava beans

MATERIALS AND METHODS

The present study was conducted in pathology department in association with department of Paediatrics, ASCOMS, Jammu. Blood samples from 100 children in various age groups presenting with anaemia and jaundice were taken up after proper counselling and consent (of parents/ guardians). Under all aseptic precautions, 3 ml of venous blood sample was drawn and dispensed in to EDTA vial for various haematological investigations. Following investigations was carried out in all patients:

- G6PD estimation by NADH reduction method. (qualitative)
- Bilirubin estimation. (Jendrassik and Grof method)
- Haemoglobin estimation. Cyanmethaemoglobin method

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RESULTS

The present study was prospective in nature conducted on 100 children in various age groups. Out of these 100 children 52 were females and 48 were males. Table 1 shows age wise distribution of children. Clinical presentation in children is depicted in table 2. Table 3 shows distribution of anemia in male and female children. Severity of anaemia and degree of jaundice is shown in table 4. Table 5 shows serum bilirubin concentration. Table 6 shows G6PD deficiency in various age groups. Table 7 shows Hb and Bilirubin in G6PD deficient children.

Table 1: Distribution of patients according to age.

S.No	Age group	No. of Patients	Percentage (%)
1	0-1 month	32	32
2	1month-1 year	30	30
3	1year-4 years	13	13
4	4years – 8 years	12	12
5	8years- 12 years	08	08
6	12years-16years	05	05

Table 2: Clinical presentation in children

S.No	Chief Complaints	No of Patients
1	Jaundice	65
2	Pallor	55
3	Fever	15
4	Generalised weakness	22
5	Chills and rigors	10
6	Pain abdomen	10

Table 3: Distribution of anaemia in male and female children

S.no	Sex	Total children	Anaemic	Total (%)
1.	Males	48	28	58.33
2.	Females	52	27	51.92

Table 4: Distribution of patients according to severity of anaemia and degree of jaundice.

S.No	Degree Of Anaemia	No of Subjects	% Age	Degree of Jaundice	No of Subjects	% Age
1	Mild (Hb 10-11.5)	20	36.36	1-6mg/dl	30	46.15
2	Moderate (7-10)	31	56.36	6-12mg/dl	26	40
3	Severe (<7)	04	07.28	>12mg/dl	09	13.85

Table 5: Distribution of Serum bilirubin concentration according to sex.

	Males	Females	Total
<1 mg/dl	15	20	35
>1mg/dl	33	32	65
Total	48	52	100

Table 6: Distribution of G6PD deficiency in various age groups.

Age groups	Total children	G6PD deficient	% of G6PD deficient children
0-1 month	32	02	6.25
1month-1 year	30	0	0
1 year – 4 years	13	01	3.03
4 years – 8 years	12	0	0
8 years – 12 years	08	0	0
12 years-16 years	05	0	0

Table 7: Distribution of Hb and Bilirubin in G6PD deficient children.

Patients	Age	Sex	Hb gm/dl	Bilirubin mg/dl
1st	4 days	Male	11	17
2nd	3 days	Male	10	26
3rd	3 years	Male	07	05

DISCUSSION

In our study a total of 3 (3%) children were found to be G6PD enzyme deficient, out of total 100 children presenting with anaemia and jaundice in various age groups. The age of presentation of children were 4 days, 3 days and 3 years, all were males. This corresponds to the study done by various authors[7,8,9]. First enzyme deficient newborn had serum bilirubin level upto 17mg% and haemoglobin 11gm%. The patient was managed by phototherapy and responded favorably by showing fall in serum bilirubin on 7th day. There was no evidence of neurological sequalae on follow up. The second enzyme deficient newborn had very high serum bilirubin exceeding 26mg% on the fifth day necessitating exchange transfusion. The baby was grossly anaemic (Hb-10gm%). The third enzyme deficient case was 3 years old presented in the OPD with chief complaints of severe pallor, respiratory distress and jaundice. After a detailed history, it was concluded that the child had ingested naphthalene balls while playing at home. Haemoglobin was 7 gm% and serum bilirubin was 5 mg%. All the 3 patients were advised regular follow up and also given the list of drugs to be avoided in future course.

Out of all age groups, G6PD deficiency was found to be most common in the age group of 0-1 month. A total of 2 (6.25%) neonates were found to be G6PD deficient out of a total of 32. This is in concordance with study by Marzban *et al.,*[10]. Indirect hyper bilirubinemia was found in all 3 G6PD deficient children, which correspond to study conducted by Atay *et al.,*[11]. The incidence of G6PD defiance in jaundiced children especially was high in this study.

CONCLUSION

To conclude, in our region G6PD deficiency is widespread in childhood especially in neonates. Screening programs help us identify those individuals who cannot be identified by routine observation and physical examination. Hence the screening for G6PD deficiency is quite essential as it can prevent complications due to the deficiency. Also it gives a clue for looking for this deficiency in the siblings of the patients.

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CITE THIS ARTICLE AS:
Purnima Gupta, Anu Gupta, Tajinder Singh. Screening of G6PD deficiency in children presenting with anaemia and jaundice. *International Journal of Bioassays* 4.11 (2015): 4508-4510.

Source of support: Nil
Conflict of interest: None Declared