

Original Research Article

Prevalence of glucose 6 phosphate dehydrogenase (G6PD) deficiency in a community by newborn screening

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Received: 04 March 2017

Revised: 30 March 2017

Accepted: 01 April 2017

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ABSTRACT

Background: Glucose 6 phosphate dehydrogenase deficiency is a genetic disorder and incidence 400 million per year globally. It is X-linked inherited disorder affect males and rarely females also by lyonisation. Characterized by significant biochemical and molecular heterogeneity. Known for its grave complications like hemolysis, severe anemia, failure and severe jaundice following ingestion of fava beans and certain drugs. Prevalent in certain communities of India, hence routine newborn screening and Detection of g6pd deficiency is important to prevent grave complications.

Methods: Prospective observational study carried out at Vani Vilas Children's hospital attached Bangalore Medical college and research institute, from January 2016 to September 2016. All the newborns born at Vani Vilas Hospital included in the study by routine newborn screening.

Results: A total of 9,136 neonates were included in this study. There were 5,013 males and 4,123 females. 37 neonates were found to be G-6-PD deficient, prevalence being 0.40%. The difference in the prevalence of G-6-PD deficiency in males 0.57% (n=29) and females 0.19% (n=8) was significant (p <0.002).

Conclusions: Significant prevalence of g6pd in India. In our study, we found 1 G6PD deficiency in per 1000 population. Hence, we recommend screening for G6PD deficiency in all the newborns to prevent complications in future.

Keywords: Deficiency, Glucose 6 phosphate dehydrogenase, Hemolysis, Heterogeneity

INTRODUCTION

Glucose-6-phosphate dehydrogenase deficiency is the most common enzyme deficiency RBCs involving more than 400 million population worldwide.¹ The exact incidence in India is not known. In India first case of G-6-PD deficiency was reported in 1961.² Glucose is the main source of energy for the red cell, which is metabolized by two major routes; the hexose monophosphate (HMP) shunt and the glycolytic pathway. G6PD is an X-linked enzyme that catalyses the first step in the HMP pathway of glucose metabolism and it

produces NADPH, which is required for the maintenance of reduced glutathione (GSH). Reduced GSH is essential for protecting red cells from oxidative damage.³ Glutathione protects RBCs from oxidative damage, deficiency is clinically manifested as neonatal hyperbilirubinemia, acute hemolytic anemia and chronic non-spherocytic hemolytic anemia.⁴ The gene for G-6-PD deficiency is located in the terminal region of the long arm of the X-chromosome at position q28. Most of the mutations affecting this gene are single base mutations. It is an X-linked condition which usually manifests itself in males carrying the mutant gene. The phenotype in females may be normal homozygote, G-6-PD deficient

homozygote or heterozygous.^{5,6} In females the condition manifests when there are two defective copies of the gene in the genome i.e., homozygous. Random X-chromosome inactivation may result in two RBC populations in female heterozygotes, one population consists of RBCs with normal G-6-PD activity and the other population with G-6-PD deficient cells.⁴ X-inactivation may be nonrandom or one or the other clone may be selected preferentially, there may be varying phenotypes and the RBCs of the heterozygous females may exhibit normal, intermediate or grossly deficient G-6-PD activity.⁷ The present study was carried out to detect the prevalence of G-6-PD deficiency and to assess the usefulness of neonatal screening for G-6-PD deficiency.

METHODS

Prospective observational study carried out at Vani Vilas Childrens hospital attached Bangalore Medical college and research institute, from January 2016 to September 2016. All the newborns born at Vani Vilas Hospital included in the study by routine newborn screening. Sample collected under aseptic precautions, 3 drops of blood on filter paper. All the sample collected after 48 hours of life and within 7 days. Sent for analysis by tandem mass spectroscopy. Babies with g6pd deficiency correlated with neonatal hyperbilirubinemia.

RESULTS

A total of 9,136 neonates were included in this study. There were 5,013 males and 4,123 females. 37 neonates were found to be G-6-PD deficient, prevalence being 0.40%. The difference in the prevalence of G-6-PD deficiency in males 0.57% (n=29) and females 0.19% (n=8) was significant (p <0.002).

Table 1: Details of cases included in the study.

	No.	%	No of positive	%
Male	5013	53.55	29	0.57
Female	4123	44.45	8	0.19
Total	9136	100	37	0.40

Incidence of hyperbilirubinemia in G-6-PD deficient Neonates, out of the 37 G-6-PD deficient neonates in this study, 11 had hyperbilirubinemia, an incidence of 29.7 %. Of these 11 neonates, two neonates required exchange transfusion. None of the neonates with hyperbilirubinemia had any other factors that could have caused hyperbilirubinemia such as ABO incompatibility, polycythemia, sepsis, in infant of diabetic mother, cephalhematoma etc.

Incidence of hyperbilirubinemia in male was significantly higher than female neonates with G-6-PD deficiency. A total of 29 male infants with G-6-PD deficiency, 9 had hyperbilirubinemia. Of 8 female neonates with G-6-PD deficiency only 2 had hyperbilirubinemia (p <0.001).

Table 2: Comparison between male and female.

	No	%
Male	29	78.37
Females	8	21.63
Total	37	100

DISCUSSION

In India, the spectrum of mutations causing glucose 6 phosphate dehydrogenase deficiency has not been well studied. However, initial studies have revealed that the G6PD Mediterranean mutation (563C→T) is the most common deficient variant followed by G6PD Kerala-Kalyan (949G→A) and G6PD Odisha (131C→G). Mediterranean G6PD was found to have significantly lower red cell enzyme activity and more severe clinical manifestations than the other two.⁸

Of the three common mutations, Odisha G6PD and Mediterranean G6PD were found to be the main mutational event causing G6PD deficiency among the tribal groups of Maharashtra, Odisha and Gujarat while Namoru (208 T→C) G6PD was exclusively found among the Dravidian speaking tribes of Nilgiri district, Tamil Nadu, which further supported the human migration from Africa to Australia along the coast of southern India.^{9,11-14} A total of 9,136 neonates were included in this study.

There were 5,013 males and 4,123 females. 37 neonates were found to be G-6-PD deficient, prevalence being 0.40%. The difference in the prevalence of G-6-PD deficiency in males % 0.57 (n=29) and females 0.19% (n=8) was significant (p <0.002). similar results found by mirtunjy et al. malay B et al.^{15,16}

We found significant difference prevalence between male and female with p value of 0.002. This is because of G6PD deficiency is an X linked disorder, the main clinical manifestations are seen in hemizygous males. In areas where G6PD deficiency is prevalent, homozygous females will also be affected in the same way as hemizygous males.

Heterozygous females can also be affected in some circumstances, because of the existence of a population of deficient cells and some cases, because of extremely skewed lyonisation, the population of deficient cells is much larger, and the total red cell enzyme concentrations may be similar to those in hemizygous males.¹⁷ Incidence of hyperbilirubinemia in G-6-PD deficient Neonates, out of the 37 G-6-PD deficient neonates in this study, 11 had hyperbilirubinemia, an incidence of 29.7 %. This is similar to study done by mirtunjy et al.

A total of 29 male infants with G-6-PD deficiency, 9 had hyperbilirubinemia. Of 8 female neonates with G-6-PD deficiency only 2 had hyperbilirubinemia (p <0.001). the p value is comparable with study done by mirtunjy et al.

CONCLUSION

Significant prevalence of g6pd in India. In our study, we found 1 G6PD deficiency in per 1000 population, 0.4% prevalent. Hence, we recommend screening for G6PD deficiency in all the newborns to prevent grave complications in future.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: The study was approved by the Institutional Ethics Committee

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Cite this article as: Moinuddin MK, Gagandeep V, Mutalik S. Prevalence of glucose 6 phosphate dehydrogenase (G6PD) deficiency in a community by newborn screening. *Int J Contemp Pediatr* 2017;4:1018-20.