

Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency In The Department Of Pediatrics At The Gabriel Toure Hospital

Diakite AA^{1,2,3*}, Traore F^{1,2,3}, Drame F^{1,2,3}, Sidibe Ln^{1,2,3}, Sacko K^{1,2,3}, Maiga B^{1,2,3}, Dicko F^{1,2,3}, Togo B^{1,2,3}, Sylla M^{1,2,3}

¹University of Sciences, Technics, and Technologies of Bamako (USTTB), Bamako-Mali.

²Faculty of Medicine and Odontostomatology, Bamako-Mali.

³Department of Pediatric of the University Hospital Center Gabriel Toure, Bamako-Mali.

*Correspondence Author:

Abdoul Aziz Diakite, Pediatric Department, Gabriel Toure Hospital/USTTB Bamako, Mali.

Received: 02 Mar 2026; **Accepted:** 06 June 2026; **Published:** 24 Sep 2026

Citation: Diakite AA, Traore F, Drame F, et al. Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency In The Department Of Pediatrics At The Gabriel Toure Hospital. *Arch Pediatr Child Health*. 2026; 1(1), 01-05.

Abstract

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common genetic enzymopathy worldwide. This study aimed to investigate the epidemiological and clinical profile of G6PD deficiency within the Department of Pediatrics at the Gabriel Touré University Hospital in Bamako, Mali. This prospective, descriptive study was conducted from December 1, 2024, to November 30, 2025. It included patients under 15 years of age who were hospitalized for anemia and jaundice, and had a laboratory-confirmed G6PD deficiency.

Twenty patients were included, representing a hospital frequency of 7.5%. Neonates aged 0-1 month constituted the majority of cases (75%). The sex ratio (M/F) was 5.6 (17 males, 3 females). Parental consanguinity was reported in 25% of cases. At admission, 90% of patients presented with jaundice ($n = 18$), and 55% had a history of blood transfusion. Indirect hyperbilirubinemia was the predominant laboratory finding. According to clinical staging, 70% of patients were classified as Stage A, 25% as Stage B, and 5% as Stage C. The mortality rate was 7.14%.

Keywords: G6PD deficiency, Jaundice, Pediatrics, Mali

Introduction

G6PD deficiency is a common X-linked genetic disorder that impairs the G6PD enzyme, which is essential for generating NADPH to shield erythrocytes from oxidative stress and subsequent hemolysis. Due to its X-linked inheritance pattern, the condition predominantly affects males. It exhibits its highest prevalence among populations of African, Mediterranean, Middle Eastern, and Southeast Asian ancestry [1, 2]. To date, more than 200 genetic variants of G6PD have been identified. Mutations within the G6PD gene significantly diminish enzymatic activity, rendering red blood cells highly susceptible to hemolysis when exposed to triggers such as fava beans, infections, oxidative stress, or specific medications, including antimalarials and sulfonamides. The World Health Organization (WHO)

categorizes these variants into classes based on residual enzyme activity and clinical severity; Classes I through III represent severe to moderate deficiency levels (<10% to 60% activity), each carrying a distinct risk of acute or chronic hemolysis [4]. Diagnosis relies on the quantitative or qualitative demonstration of reduced G6PD enzyme activity, whereas molecular genetic testing is rarely required for routine clinical management. Consequently, patient management remains primarily preventive, focusing heavily on the strict avoidance of fava beans, oxidative medications, and other known hemolytic triggers [5].

Methodology

This prospective, descriptive study was conducted from December 1, 2024, to November 30, 2025, within the

Department of Pediatrics at the Gabriel Touré University Hospital in Bamako, Mali. The study enrolled patients under 15 years of age who met the following criteria: Hospitalization within the pediatric department during the study period, presentation with concurrent anemia and jaundice and laboratory-confirmed glucose G6PD deficiency.

Data were gathered using a standardized abstraction form. Information was extracted from comprehensive medical records, real-time clinical examinations, and structured interviews with the patients and/or their parents or legal guardians. The analyzed variables encompassed sociodemographic characteristics and clinical presentations. According to our reference laboratory, the normal range for G6PD enzymatic activity was defined as 8.0–14.9 U/g Hb. Values strictly below 8.0 U/g Hb were classified as G6PD deficiency, in alignment with the updated 2024 World Health Organization (WHO) classification criteria.

In neonates, total hyperbilirubinemia thresholds were defined chronologically based on postnatal age as follows: <24 hours: >100 µmol/L, 24–48 hours: >170 µmol/L, 48–72 hours: >205 µmol/L and >72 hours: >257 µmol/L. For older infants and pediatric patients, hyperbilirubinemia was defined as a total serum bilirubin level exceeding 70 µmol/L.

Results

Twenty patients were enrolled (n = 20), representing an overall hospital prevalence of 7.5%. The cohort demonstrated a strong neonatal predominance, with infants aged 0-1 month accounting for 75.0% of cases. There was a notable male bias, with a sex ratio (M/F) of 5.6 (17 males and 3 females). Regarding familial and genetic background, 10 % of patients had siblings with a documented history of G6PD deficiency, and parental consanguinity was reported in 25.0% of cases. At the time of admission, clinical jaundice was the most common presentation, observed in 90.0% of patients (n = 18). Over half of the study population (55.0%) had a history of blood transfusion, and 60.0% (n

= 12) presented with anemia. Concomitant infection was identified as the most frequent comorbidity, affecting 45.0% of the cohort (n = 9). Unconjugated hyperbilirubinemia was the predominant laboratory abnormality, present in 70.0% of patients (n = 14). Within this biological subgroup, indirect bilirubin levels were severely elevated in 93.0% of cases (n = 13). The distribution of ABO and Rh blood groups among the patients was as follows: O positive: 40.0% (n = 8), A positive: 25.0% (n = 5) and B positive: 20.0% (n = 4). According to clinical and biological staging, 70.0% of patients (n = 14) were classified as Stage A, 25.0% (n = 5) as Stage B and 5.0% (n = 1) as Stage C. The overall in-hospital mortality rate was 5.0% (n = 1).

Discussion

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is recognized as one of the most prevalent inherited enzymopathies globally, presenting a significant clinical burden among pediatric populations across sub-Saharan Africa [6]. This enzymopathy impairs erythrocyte integrity, predisposing patients to acute hemolytic anemia upon exposure to specific oxidative triggers, including infections, fava beans, or specialized pharmacotherapies [7].

The epidemiological landscape of G6PD deficiency exhibits substantial geographic and ethnic heterogeneity. In African pediatric cohorts, the reported prevalence rates vary widely, ranging from under 1.0% to nearly 24.0% depending on the region and diagnostic methodology. Due to its X-linked recessive mode of inheritance, a distinct male predominance is systematically documented across literature. For instance, in Nigeria, epidemiologic data indicate a prevalence of approximately 15.0% among children, with a significantly higher burden observed in males. Comparable demographic trends and prevalence variations have been documented in neighboring and regional nations, including Burkina Faso, Ghana, Kenya, Gabon, and the Democratic Republic of Congo, which aligns with the high male-to-female sex ratio observed in our study [9], (Table 1).

Characteristics	Number	Percent
Age of patients		
< 1 month	15	75
1 -12 month	2	10
> 12 month	3	15
Gender of patients		
Female	3	15
Male	17	85
Consanguinity		
yes	5	25
No	15	75
G6PD background		
Yes	2	10
No	18	90

Discovery circumstances		
jaundice	18	90
Pallor	2	10
G6PD levels		
< 3	14	70
[3-6,74]	5	25
[6,75-8]	1	5
Classification G6PD deficiency		
Classe A	14	70
Classe B	5	25
Classe C	1	5

Table 1: Patient characteristics

The G6PD A-variant represents the predominant genetic polymorphism identified within African populations. This specific variant underscores the critical clinical necessity for routine targeted screening, particularly prior to the administration of oxidant medications [10]. Crucially, in malaria-endemic regions like Mali, pre-treatment screening is mandatory before deploying 8-aminoquinoline antimalarials such as primaquine or tafenoquine to mitigate the risk of severe, potentially fatal drug-induced hemolytic crises. Although the G6PD A-variant is traditionally classified as a moderate (Class III) deficiency, it remains a potent driver of acute hemolytic crises during active infections or following exposure to oxidizing agents [11]. Despite the high disease burden among pediatric populations, routine neonatal screening for G6PD deficiency remains severely constrained across sub-Saharan Africa due to healthcare infrastructure deficits and financial limitations. Consequently, the vast majority of pediatric patients are diagnosed only after presenting with clinical symptoms. In our series, the genetic architecture of this cohort was analyzed to map transmission dynamics across maternal and paternal lineages [12,13]. This evaluation underscored the pivotal role of parental consanguinity and familial clustering among siblings. Crucially, most G6PD-deficient children remain asymptomatic and clinically unremarkable until exposed to an exogenous oxidative trigger [14]. Once triggered, accelerated erythrocyte destruction outpaces bone marrow compensatory capacity, precipitating acute hemolytic anemia [15]. In neonates, the primary clinical manifestation of this hemolytic process is severe jaundice [16]. Delayed recognition or inadequate management of this hyperbilirubinemia poses a profound risk of acute bilirubin encephalopathy or kernicterus. Epidemiological data across Africa consistently reaffirm this strong pathogenic link between G6PD deficiency and neonatal jaundice. Notably, recent systematic reviews and meta-analyses indicate that nearly 25.0% of all jaundiced African neonates harbor an underlying G6PD deficiency, a finding that mirrors the high rate of clinical jaundice (90.0%) and neonatal predominance observed in our study population [17]. During an acute hemolytic episode, patients typically present with standard signs of severe anemia, including prominent pallor, lethargy,

and generalized asthenia. Compensatory cardiovascular and respiratory responses frequently manifest as tachycardia and tachypnea. Intravascular hemolysis triggers the excretion of free hemoglobin into the urinary tract, producing a pathognomonic dark, tea- or cola-colored urine. Additional clinical signs include irritability, dizziness, moderate pyrexia, and localized abdominal or lumbar pain.

The clinical spectrum varies markedly among pediatric patients. While some children experience mild, self-limiting episodes that resolve spontaneously upon the removal of the offending agent, others develop life-threatening anemia requiring immediate hospitalization and supportive blood transfusions [18]. The primary pathogenic triggers identified in this pediatric population include *Plasmodium falciparum* malaria infections, fava bean ingestion (favism), exposure to naphthalene-containing mothballs, and specific oxidizing medications—predominantly sulfonamide antibiotics and 8-aminoquinoline antimalarials. Consequently, early recognition of these clinical features and strict avoidance of known oxidative stressors remain the cornerstone of effective patient management. Biochemically, G6PD deficiency is characteristically unmasked during active hemolytic crises when erythrocyte destruction accelerates dramatically. The primary hematological hallmark is a precipitous drop in hemoglobin levels, corroborating acute hemolytic anemia [19].

Complete blood counts (CBC) during acute hemolytic episodes characteristically reveal a precipitous decline in total erythrocyte counts alongside markedly reduced hematocrit levels. In response to accelerated peripheral erythrocyte destruction, the bone marrow initiates compensatory erythropoiesis, which is biologically manifested by a pronounced increase in the absolute reticulocyte count. Concurrently, a suite of classic biochemical markers unmasks the hemolytic process. Accelerated red cell lysis leads to a substantial elevation in total serum bilirubin, driven predominantly by the unconjugated (indirect) fraction. Intracellular enzyme leakage causes a marked rise in serum lactate dehydrogenase (LDH) levels, while scavenging mechanisms free plasma hemoglobin result in a significant depletion of serum haptoglobin. In cases of severe,

uncontrolled intravascular hemolysis, the renal threshold for hemoglobin reabsorption is exceeded, precipitating macroscopically visible hemoglobinuria. While these indirect bio-indicators are highly suggestive of a hemolytic event, a definitive diagnosis of G6PD deficiency requires the direct quantitative or qualitative measurement of G6PD enzymatic activity within the blood. Affected pediatric patients characteristically exhibit residual enzyme activity levels significantly below established laboratory reference intervals. Integrating these hematological and biochemical profiles is clinically paramount, as they provide healthcare professionals with the objective metrics required for early diagnosis, risk stratification, and real-time therapeutic monitoring during active hemolytic crises [20].

Conclusion

G6PD deficiency is a highly prevalent hereditary enzymopathy that remains systematically underdiagnosed within pediatric healthcare settings in Mali. This study underscores that diagnostic identification is fully achievable as early as the neonatal period through targeted enzymatic activity testing. To mitigate the significant morbidity, potential neurological sequelae, and mortality associated with this genetic disorder, the implementation of universal or targeted routine neonatal screening programs is critically required across the national healthcare infrastructure.

Acknowledgements

We would like to thank all the patients and their families for agreeing to participate in this study and for allowing the publication of these data.

Authors' contributions

Traoré Fousseyni wrote the original manuscript text and conducted conceptualization, study design, methodology, and interpretation of the data. Abdoul aziz Diakite conducted the data collection, formal data analysis and the interpretation of the data. All authors revised the manuscript and approved the final version to be published.

Funding

This research study did not receive any grant from any funding agency.

Data availability

The database analyzed during the current study is available from the corresponding author.

Declarations

Ethics approval and consent to participate

Upon admission, patients' parents or legal guardians approved their enrolment in a clinical research project in accordance with the national ethics committee of Mali. The national ethics committee of Mali has approved the research protocol. The registration number is 002632 of national ethics committee of Mali.

Consent for publication

Not applicable.

Conflicts of interest: the authors have not declared any conflicts of interest.

References

1. Harcke SJ, Rizzolo D, Harcke HT. G6PD deficiency: An update. *JAAPA*. 2019;32(11):21-26. doi: 10.1097/01.JAA.0000586304.65429.a7.
2. Beutler E. G6PD deficiency. *Blood*. 1994;84(11):3613-36.
3. Belfield KD, Tichy EM. Review and drug therapy implications of glucose-6-phosphate dehydrogenase deficiency. *Am J Health Syst Pharm*. 2018;75(3):97-104. doi: 10.2146/ajhp160961.
4. Cappellini MD, Fiorelli G. Glucose-6-phosphate dehydrogenase deficiency. *Lancet*. 2008;371(9606):64-74. doi: 10.1016/S0140-6736(08)60073-2.
5. Frank JE. Diagnosis and management of G6PD deficiency. *Am Fam Physician*. 2005;72(7):1277-82.
6. Luzzatto L, Nannelli C, Notaro R. Glucose-6-Phosphate Dehydrogenase Deficiency. *Hematol Oncol Clin North Am*. 2016;30(2):373-93. doi: 10.1016/j.hoc.2015.11.006.
7. Doshi BS, Kamdar A, Lambert MP, Obstfeld AE. Hemolysis After Medication Exposure in Pediatric Patients With G6PD Deficiency. *J Pediatr Hematol Oncol*. 2022;44(5):261-263. doi: 10.1097/MPH.0000000000002350.
8. Williams O, Gbadero D, Edowhorhu G, Brearley A, Slusher T, Lund TC. Glucose-6-phosphate dehydrogenase deficiency in Nigerian children. *PLoS One*. 2013;8(7):e68800. doi: 10.1371/journal.pone.0068800.
9. Taleb M, Brahim AT, Yebouk C, Khairy T, Ishagh B, Lyoussi B, Ghaber SM. Epidemiology of glucose-6-phosphate dehydrogenase deficiency in 10 malaria-endemic countries in sub-Saharan Africa: a systematic review. *Pan Afr Med J*. 2025;50:7. doi: 10.11604/pamj.2025.50.7.45066.
10. Howes RE, Battle KE, Satyagraha AW, Baird JK, Hay SI. G6PD deficiency: global distribution, genetic variants and primaquine therapy. *Adv Parasitol*. 2013;81:133-201. doi: 10.1016/B978-0-12-407826-0.00004-7.
11. Batista da Rocha J, Othman H, Hazelhurst S. Molecular dynamics of G6PD variants from sub-Saharan Africa. *Biochem Biophys Rep*. 2022;30:101236. doi: 10.1016/j.bbrep.2022.101236.
12. Mohamed GS, Lemine SM, Cheibetta S, Mohamed A. Dépistage néonatal du déficit en glucose 6 phosphate déshydrogénase (G6PD) en Mauritanie [Neonatal screening for glucose-6-phosphate dehydrogenase (G6PD) deficiency in Mauritania]. *Pan Afr Med J*. 2018;30:224. doi: 10.11604/pamj.2018.30.224.13947.
13. Orubu E, Satrom K, Ezenwa B, Fajolu I, Lund T, Obi A, Ezeaka C, Slusher T. Real-world use of the Carestart™ glucose-6-phosphate dehydrogenase rapid diagnostic test to determine G6PD deficiency in Nigerian neonates. *J Trop Pediatr*. 2024;70(6):fmae050. doi: 10.1093/tropej/fmae050
14. Aynalem YA, Mulu GB, Akalu TY, Shiferaw WS. Prevalence of neonatal hyperbilirubinaemia and its association with glucose-6-phosphate dehydrogenase deficiency

-
- and blood-type incompatibility in sub-Saharan Africa: a systematic review and meta-analysis. *BMJ Paediatr Open*. 2020;4(1):e000750. doi: 10.1136/bmjpo-2020-000750.
15. Mehta A, Mason PJ, Vulliamy TJ. Glucose-6-phosphate dehydrogenase deficiency. *Baillieres Best Pract Res Clin Haematol*. 2000;13(1):21-38. doi: 10.1053/beha.1999.0055.
16. Lee HY, Ithnin A, Azma RZ, Othman A, Salvador A, Cheah FC. Glucose-6-Phosphate Dehydrogenase Deficiency and Neonatal Hyperbilirubinemia: Insights on Pathophysiology, Diagnosis, and Gene Variants in Disease Heterogeneity. *Front Pediatr*. 2022;10:875877. doi: 10.3389/fped.2022.875877.
17. Kassahun W, Tunta A, Abera A, Shiferaw M. Glucose-6-phosphate dehydrogenase deficiency among neonates with jaundice in Africa; systematic review and meta-analysis. *Heliyon*. 2023;9(7):e18437. doi: 10.1016/j.heliyon.2023.e18437.
18. Youngster I, Arcavi L, Schechmaster R, Akayzen Y, Popliski H, Shimonov J, Beig S, Berkovitch M. Medications and glucose-6-phosphate dehydrogenase deficiency: an evidence-based review. *Drug Saf*. 2010;33(9):713-26. doi: 10.2165/11536520-000000000-00000.
19. Milburn S, Bhutani VK, Weintraub A, Guttmann K. Implementation of Universal Screening for G6PD Deficiency in Newborns. *Pediatrics*. 2024;154(2):e2024065900. doi: 10.1542/peds.2024-065900.
20. DelFavero JJ, Jnah AJ, Newberry D. Glucose-6-Phosphate Dehydrogenase Deficiency and the Benefits of Early Screening. *Neonatal Netw*. 2020;39(5):270-282. doi: 10.1891/0730-0832.39.5.270.

Copyright:

©2026 Diakite AA, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution 4.0 International License.