Case study

EARLY DETECTION AND MANAGEMENT OF GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY THROUGH NEWBORN SCREENING: A CASE STUDY AND PUBLIC HEALTH PERSPECTIVE

ABSTRACT:

Newborn screening is a crucial public health program aimed at early detection of genetic, metabolic and endocrine disorders in infants, enabling prompt intervention to prevent long-term complications and improve outcomes. This study focuses on the importance of screening for conditions such as Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency a hereditary enzyme disorder that can cause life-threatening hemolysis. The case study of a 3-day-old infant diagnosed with G6PD deficiency highlights the significance of early diagnosis through newborn screening. Following diagnosis, parental counseling, trigger avoidance and vaccination were emphasized to manage the condition effectively. The study advocates for the integration of G6PD screening into routine neonatal screening programs particularly in regions with high endemicity to ensure timely interventions, reduce morbidity and enhance quality of life. The findings underscore the critical role of early detection, caregiver education and healthcare policy integration in improving outcomes for infants with genetic and metabolic disorders.

Keywords: Newborn screening, Glucose-6-Phosphate Dehydrogenase deficiency, Metabolic disorders.

INTRODUCTION

Newborn screening is a vital public health program designed to detect genetic, metabolic, hormonal and functional disorders in infants shortly after birth enabling timely interventions to prevent severe complications improve long-term outcomes and in some cases save lives. Typically performed within the first few days of life it involves collecting a small blood sample

from the baby's heel to analyze for various conditions based on the country's screening panel [1]. Many programs also include additional tests such as hearing screenings for congenital hearing loss and pulse oximetry to identify critical congenital heart defects [2]. These screenings are crucial for detecting conditions that may not present symptoms at birth but can cause irreversible damage or life-threatening complications if untreated [3]. The inclusion of disorders like metabolic, endocrine and immunological conditions ensures comprehensive health evaluation while advancements in diagnostic technologies such as tandem mass spectrometry have expanded the range of detectable conditions [4]. Beyond medical benefits newborn screening reduces long-term healthcare costs by preventing severe disabilities and chronic illnesses, while empowering parents and healthcare providers with essential information to improve the child's quality of life. As a cornerstone of preventive medicine newborn screening underscores the importance of early detection in building a healthier future generation [5].

Neonatal TSH (Thyroid-Stimulating Hormone):

Neonatal TSH levels are measured to screen for congenital hypothyroidism, a condition where the thyroid gland does not produce enough thyroid hormone. Early detection through TSH screening can prevent developmental delays and intellectual disabilities by initiating thyroid hormone replacement therapy shortly after birth [6].

17 OHP (17-Hydroxyprogesterone):

Elevated levels of 17 OHP can indicate congenital adrenal hyperplasia (CAH), a group of inherited disorders that affect adrenal gland function. CAH can lead to severe imbalances in hormone production, resulting in potentially life-threatening electrolyte disturbances and sexual development issues. Early detection allows for prompt treatmentreducing the risk of adrenal crisis [7].

Immunoreactive Trypsin (IRT):

The IRT test is used to screen for cystic fibrosis (CF), a genetic disorder that affects the lungs, pancreas and other organs. IRT levels are elevated in newborns with CF due to the impaired function of the pancreas. A positive IRT result prompts further genetic testing, enabling early intervention to manage CF and improve the infant's long-term health [8].

Total Galactose:

Screening for total galactose levels is part of the testing for galactosemia an inherited metabolic disorder that affects the body's ability to process galactose, a sugar found in milk. Elevated galactose levels can lead to liver damage, cataracts and brain damage if untreated. Early diagnosis and dietary modifications can prevent severe complications and developmental issues in affected infants [9].

Glucose-6-Phosphate Dehydrogenase (G6PD):

The G6PD test screens for glucose-6-phosphate dehydrogenase deficiency, an X-linked enzyme disorder that can cause hemolytic anemia. This condition can be triggered by certain infections or medications leading to the destruction of red blood cells. Early identification allows for the avoidance of triggering factors and management of symptoms preventing potentially severe complications [10].

CASE:

3-day-old male infant born via normal delivery with no complications during pregnancy or delivery underwent routine newborn screening on day 3. The screening results showed negative

Test Name	Observed Value	Units	Biological Reference Interval	Interpretation
Neonatal TSH Method: TRFIA*	< 0.7	µU/ml	Negative : < 10 µU/ml Positive: >= 10 µU/ml	Screen Negative
Neonatal 17 OHP Method: TRFIA*	< 5.3	nmol/L	Full Term: Negative < 20 Positive > = 20 Pre Term (Weight < 2500g): Negative < 30 Positive > = 30 Pre Term (Weight >= 2500g): Negative < 20 Positive > = 20	Screen Negative
Neonatal Immunoreactive Trypsin Method: TRFIA*	31.9	ng/mL	Negative : < 70 Positive : > = 70	Screen Negative
Neonatal Total Galactose Method: Oxidase	3.96	mg/dl	Negative : < 10 Positive: > = 10	Screen Negative
Glucose-6-phosphate dehydrogenase Method: Oxidase	0.6	U/g Hb	Negative : > 2.5 Positive : < = 2.5	Screen Positive Condition : G6PD Deficiency
*TRFIA - Time Resolved FluoroImmuno Assay.				

Table 1: shows the observed value of G6PD, screen positive condition

results for Neonatal TSH, 17-OHP, Immunoreactive Trypsin (IRT), Total Galactose and a Positive result for G6PD. Confirmatory testing revealed a deficient G6PD value of 1.04 U/g Hb (normal range: 6.0–10.5 U/g Hb), leading to a diagnosis of Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency. This hereditary X-linked recessive disorder impairs the red blood cells ability to handle oxidative stress increasing the risk of hemolysis in response to certain triggers such as infections, medications or specific foods.

G6PD Mechanism:

Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency is an X-linked genetic disorder affecting the G6PD enzyme which plays a critical role in the pentose phosphate pathway by generating NADPH to maintain reduced glutathione (GSH). This deficiency impairs the ability of red blood cells (RBCs) to neutralize reactive oxygen species (ROS), leading to oxidative stress, hemoglobin oxidation and the formation of Heinz bodies. These changes make RBCs prone to hemolysis resulting in acute hemolytic anemia characterized by jaundice, fatigue and hemoglobinuria particularly under oxidative stress[11].

Parental Counselling:

The baby's clinical course involved immediate interventions, starting with parental counseling to educate the parents about G6PD deficiency, its implications and the importance of avoiding triggers such as certain medications, fava beans and naphthalene-containing mothballs. The baby was closely monitored for signs of hemolysis, such as jaundice, pallor and dark-colored urine with regular blood tests scheduled to assess bilirubin levels, hemoglobin and reticulocyte counts to track the baby's condition.

Investigation	Observed Value		Biological Reference Interval
G6PD test by kinetic method	16.00	units/dl of blood	
G6PD-quantitative, blood by Kinetic method	Deficient,1.04	Units/gm of HB	7.29-10.36

Table 2:display the observed value of G6PD quantitative by kinetic method

Long-term Management:

For long-term management parents were advised to avoid known oxidative stressors and remain vigilant to minimize potential triggers [12]. Timely vaccinations were emphasized to reduce the

risk of infections, as even minor infections could potentially worsen the condition due to heightened oxidative stress. Vaccines such as pneumococcal, influenza and meningococcal were specifically highlighted for their role in preventing severe bacterial and viral infections [13]. Parents were educated on the importance of adhering to the vaccination schedule and informed about booster doses to maintain long-term immunity [14]. Additionally, they were provided with contact information for emergency healthcare access to ensure preparedness in case of any urgent situations [15].

DISCUSSION:

The significance of early detection through newborn screening cannot be overstated especially for conditions like glucose-6-phosphate dehydrogenase (G6PD) deficiency where timely intervention can prevent potentially life-threatening hemolytic crises. Screening at birth enables healthcare providers to identify at-risk infants early and implement proactive management strategies, reducing morbidity and improving long-term outcomes. This case underscores the critical need to integrate G6PD testing into routine neonatal screening programs, particularly in regions with a high prevalence of the disorder. Such programs not only facilitate early diagnosis but also provide an opportunity to educate families about avoiding known triggers like certain foods, medications and infections [16][17].

The parental role in management is crucial as caregivers are the first line of defense in recognizing symptoms and seeking timely medical attention. Educating parents on preventive measures, symptom recognition and the importance of timely vaccinations is essential for reducing infections a common trigger for hemolysis. Providing emergency contact information and practical guidance further equips families to manage potential crises effectively [18].

Incorporating G6PD screening into national healthcare policies would be a significant public health milestone especially in regions with high endemicity for malaria or hemoglobinopathies where the prevalence of G6PD deficiency is elevated. Collaboration between healthcare systems and policymakers can ensure accessibility to screening programs and equitable care. Overall the combination of early detection, caregiver education and systemic healthcare improvements can significantly reduce complications and enhance the quality of life for affected individuals [19][20].

CONCLUSION:

This study highlights the vital role of newborn screening in early identification of conditions such as Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency, which if undiagnosed can lead to life-threatening hemolytic crises. Early detection through routine screening enables timely interventions, preventing severe complications and improving long-term outcomes. The case study of the 3-day-old infant with G6PD deficiency demonstrates the importance of parental counseling, lifestyle adjustments and vigilant monitoring in managing the condition. By educating families on trigger avoidance the importance of vaccinations and emergency preparedness the risk of exacerbating the disorder can be minimized. Additionally, integrating G6PD screening into national healthcare policies particularly in regions with high prevalence is essential for ensuring equitable access to care and reducing the incidence of preventable health complications. The study emphasizes that early detection, education and a collaborative healthcare approach are key components in improving the health and quality of life for affected infants.

COMPETING INTERESTS DISCLAIMER:

Authors have declared that they have no known competing financial interests OR non-financial interests OR personal relationships that could have appeared to influence the work reported in this paper.

DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Author(s) hereby declares that NO generative AI technologies such as Large Language Models (ChatGPT, COPILOT, etc.) and text-to-image generators have been used during the writing or editing of this manuscript.

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