A-249. Preliminary Investigation into the Prevalence of G6PD Deficiency in a Pediatric African American Population using a Near-Patient Diagnostic Platform

[1]Van Leung-Pineda, [1]Elizabeth Weinzierl, [1]Beverly Rogers, [2]Ramakrishna Sista
[1] Department of Pathology and Laboratory Medicine, Children's Healthcare of Atlanta, Emory University School of Medicine, Atlanta, GA, USA
[2] Baebies, Inc., Durham, NC, USA

BACKGROUND

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common enzyme deficiency in humans, affecting 400 million people worldwide with the vast majority of those being asymptomatic and undiagnosed. It has a high prevalence in persons of African, Asian, and Mediterranean descent. G6PD deficiency results from mutations in the G6PD gene, and is inherited as an X-linked recessive disorder. G6PD deficiency is polymorphic, with more than 300 known variants. The enzyme protects red blood cells from the effects of potentially harmful molecules called reactive oxygen species which are byproducts of normal cellular functions. Chemical reactions involving glucose-6-phosphate dehydrogenase produce compounds that prevent reactive oxygen species from building up to toxic levels within red blood cells. Therefore G6PD deficient patients are at a disproportionate risk for developing hemolytic anemia and subsequent hyperbilirubinemia.

In addition, jaundiced patients who are also G6PD deficient are less likely to respond well to conventional treatment methods and are at greater risk for suffering from the long-term consequences of neonatal hyperbilirubinemia. Additionally, G6PD deficient patients remain susceptible to hemolytic anemia throughout the course of their lives, after exposure to certain viral or bacterial pathogens or treatment with common medications. Early diagnosis of G6PD deficiency is vital to preventing serious complications later on in life. In the last 3 years, an average of 100 G6PD deficiency tests were performed per year for Children's Healthcare of Atlanta's patient population. We hypothesize that the incidence of G6PD might be larger than thought based on those testing volumes.

Baebies previously developed a low blood volume, quantitative test for G6PD on a digital microfluidic (DMF) platform to rapidly analyze whole blood samples near the patient in a clinical setting.

OBJECTIVE

The primary objective of this study was to establish the prevalence of G6PD deficiency among African American newborns and pediatric subjects at Children's Healthcare of Atlanta, by randomly testing residual laboratory samples. A secondary objective was to evaluate the use of near patient diagnostic platform in a clinical core laboratory setting

METHODOLOGY

We performed a retrospective study of residual, de-identified whole blood specimens collected at the Children's Healthcare of Atlanta-Egleston Campus to study the prevalence of G6PD deficiency in the African-American population. Residual and de-identified whole blood specimens (n=152) were obtained from pediatric African American patients under an IRB approved protocol. Samples were stripped of patient identifiers prior to testing on the DMF platform. Only race, ethnicity, liver enzyme test results when available (obtained by standard- laboratory methods), date of collection, age, and gender information were collected specific to this study. Specimens were selected randomly from residual samples available in the lab. Samples were analyzed on the DMF G6PD platform within 48 hours of sample collection at CHOA to obtain G6PD values (U/gHb).

Fig. 1) Baebies Finder Testing platform for detecting G6PD Deficiency in blood

RESULTS

PATIENT DEMOGRAPHICS	
N All	152
Median Age All	11.3 Y
Low Age All	17 d
High Age All	20 Y
N Female	78
Median Age Female	12 Y
Low Age Female	17 d
High Age Female	20 Y
N Male	74
Median Age Male	10.5 Y
Low Age Male	2.1 m
High Age Male	19 Y

Table I) Study Cohort Demographic Characteristics

G6PD Activity Results			
Parameter	**All**	**Female**	**Male**
ALL			
Median G6PD Value (U/gHb)	10.5	10.6	10.3
Mean G6PD Value (U/gHb)	10.0	10.7	9.3
SD of G6PD Values (U/gHb)	4.0	3.5	4.3
Deficient Samples (N)	14	3	11
Deficiency Frequency	9.21%	3.85%	14.86%
NORMAL			
Median G6PD Value (U/gHb)	10.8	10.7	10.8
Mean G6PD Value (U/gHb)	10.9	11.0	10.7
SD of G6PD Values (U/gHb)	3.0	3.1	2.9
DEFICIENT			
Median G6PD Value (U/gHb)	1.4	1.3	1.5
Mean G6PD Value (U/gHb)	1.3	1.5	1.3
SD of G6PD Values (U/gHb)	0.5	0.4	0.5

Table II) G6PD Activity Results From Pediatric Residual Samples
G6PD Activity < 3.5 U/gHb is considered deficient

CONCLUSION

The near-patient DMF G6PD assay was used to estimate the prevalence of G6PDD deficiency in a pediatric African American cohort. The overall prevalence of deficiency was found to be 9.21%, consistent with what is reported in the literature (1). The incidence of G6PD deficiency in males (14.83%) was greater than in females (3.85%), also consistent with previous reports.

Our results suggest that there may be a significant population of African American patients, especially male, who may not be known to be G6PD deficient, and that this condition may not be considered when medical decisions are being made about their care. A future study is planned to correlate whether G6PD deficiency in this population is associated with abnormal liver function tests.

REFERENCE

1. Chinevere TD et al. Prevalence of glucose-6-phosphate dehydrogenase deficiency in U.S. Army personnel. Mil Med. 2006 (9) 905-7