

**Gene mutation and qualitative enzyme assay of G6PD
enzyme deficiency in neonatal hyperbilirubinemia**

Thesis

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Contents

	pages
• List of tables	I
• List of figures	IV
• List of abbreviations	VI
• Aim of work	VIII
• <u>Review of literature:</u>	
• Introduction	1
• Chapter 1: Neonatal Hyperbilirubinemia	3
• Chapter 2: Glucose 6 Phosphate Dehydrogenase Deficiency.....	34
• Patients and methods	71
• Results	90
• Discussion	112
• Conclusion and Recommendations	126
• Summary	128
• Appendix	130
• References	135
• Arabic summary	i

AIM OF WORK

Abstract

Background: Molecular characterization of glucose-6-phosphate dehydrogenase (G6PD) deficiency variants is essential, especially since the biochemical characterization has lost its significance due to the individual variability. As a result, cases can be misdiagnosed. The present study was designed to determine the incidence of G6PD Mediterranean (Med) mutation among Egyptian children with G6PD deficiency as well as its molecular association with the G6PD 1311T silent polymorphism.

Methods: Fifty full term male, Egyptian neonates, presented with neonatal hyperbilirubinemia were subjected to quantitative G6PD enzyme assay. A polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) was used for detection of G6PD Mediterranean mutation and G6PD 1311 silent polymorphism.

Results: Qualitative assay of G6PD level revealed presence of 21 cases (42%) out of 50 cases with neonatal hyperbilirubinemia had G6PD deficiency. Within the G6PD deficient group, there were 10 cases (47.62%) had mild G6PD deficiency, 4 cases (19.05%) had moderate G6PD deficiency, and 7 cases (33.33%) had severe G6PD deficiency. In this study, all cases with gene mutation were G6PD deficient, where 33.33% (7/21) cases had G6PD Mediterranean mutation (563 C-T), and 28.57% (6/21) cases had G6PD Silent mutation at Position (1311 C-T).

Conclusion: G6PD deficiency seems to be a relatively common cause of neonatal jaundice in Egyptian infants. Detection of this enzymopathy by measuring G6PD level and molecular analysis of gene mutations can give a clue for early diagnosis of G6PD deficiency, monitoring for possible jaundice, and consequent prevention of possible complication as kernicterus in neonatal period, and hemolytic crisis later on.

Key words: G6PD deficiency, Mediterranean mutation, PCR-RFLP, 1311 silent polymorphism.

List of Tables

No	Title	Page
1-	Risk factors for development of severe hyperbilirubinemia in infants ≥ 35 weeks of gestation.....	4
2-	Non Conjugated hyperbilirubinemia	5
3-	Conjugated hyperbilirubinemia	6
4-	Differential diagnosis of Neonatal Jaundice	7
5-	Different types of devices used for transcutaneous bilirubin measurement	18
6-	Laboratory Evaluation of the Jaundiced Infant of 35 or More Weeks' gestation.....	20
7-	Postdischarge and follow up based on predischage TSB (mg/dl) level (based on hour-specific bilirubin nomogram).....	33
8-	Classes of G6PD deficiency.....	42
9-	Comparison of the Two Most Common Variants of G6PD Deficiency.....	44
10-	Drugs that may cause hemolysis in G6PD deficient patients with popolymorphic variants.....	51
11-	Factors that affect individual susceptibility to, and severity of drug-induced oxidative hemolysis.....	52
12-	Drugs and chemicals associated with substantial hemolysis in patients with G6PD deficiency.....	56
13-	Drugs that can be probably safe to be given in normal therapeutic doses to G6PD deficient individuals, but not in those with non-spherocytic hemolytic anemia.....	68
14-	Demographic data of the studied cases.....	91
15-	Clinical and Laboratory finding in G6PD deficient versus G6PD normal cases	92

16-	Relationship between 1311 silent polymorphism and postnatal age at presentation, gestational age, birth weight, and total serum bilirubin.....	93
17-	Correlation between Mediterranean 563 C/T mutation and postnatal age at presentation, gestational age, birth weight, and total serum bilirubin.....	94
18-	Mode of delivery and 1311 silent polymorphism.....	95
19-	Mode of delivery and Mediterranean 563 C/T mutation.....	96
20-	Hemoglobin level and 1311 silent polymorphism.....	96
21-	Relationship between hemoglobin (HGB) level and G6PD enzyme assay.....	97
22-	Reticulocytic count and 1311 silent polymorphism.....	98
23-	Relationship between reticulocytic count and G6PD enzyme assay.....	99
24-	Albumin and 1311 silent polymorphism.....	99
25-	Albumin and Mediterranean 563 C/T mutation	100
26-	Relationship between albumin level and G6PD enzyme assay..	100
27-	Treatment and Mediterranean 563 C/T mutation	101
28-	Relationship between treatment and G6PD enzyme assay....	102
29-	Correlation between complications and G6PD enzyme assay...	105
30-	G6PD enzyme assay and 1311 silent polymorphism.....	105
31-	G6PD enzyme assay and Mediterranean 563 C/T mutation.....	106
32-	Relationship between age and TSB, and G6PD enzyme assay..	106
33-	Bilirubin/Albumin ratio in G6PD deficient cases.....	108
34-	Bilirubin/Albumin ratio in 1311 silent polymorphism cases.....	109
35-	Bilirubin/Albumin ratio in 563 Mediterranean gene mutation cases.....	109
36-	Correlation between Total serum bilirubin and hemoglobin, reticulocytic count, and B/A ratio in G6PD deficient cases.....	110

37-	Correlation between Total serum bilirubin and hemoglobin, reticulocytic count, and B/A ratio in 1311 silent polymorphism cases.....	110
38-	Correlation between Total serum bilirubin and hemoglobin, reticulocytic count, and B/A ratio in 563 Mediterranean gene mutation cases.....	111

List of Figures

No	Title	Page
1-	Bilirubin metabolism.....	9
2-	Schematic representation of the steps involved in bilirubin (B) throughput in hepatocytes.....	10
3-	Hour-specific percentile risk for emergence of significant hyperbilirubinemia in healthy term and near-term infants.....	16
4-	Guidelines for exchange transfusion in infants 35 or more weeks' gestation	28
5-	Algorithm for management of neonatal jaundice in nursery	29
6-	Algorithm providing recommendations for management and follow-up according to predischage bilirubin measurements, gestation, and risk factors for subsequent hyperbilirubinemia	30
7-	Nomogram for designation of risk in 2840 well newborns at ≥ 36 weeks' gestational age with birth weight of ≥ 2000 g or ≥ 35 weeks' gestational age and birth weight of ≥ 2500 g based on the hour-specific serum bilirubin values	31
8-	Pentose phosphate pathway	35
9-	Genotype-phenotype associations in the G6PD tetrameric structure.....	36
10-	The amino acid concentration of G6PD enzyme	37
11-	Location of <i>G6PD</i> gene on X chromosome	38
12-	Most common mutations along coding sequence of <i>G6PD</i> gene.....	39
13-	Genotype–phenotype heterogeneity in the pathogenesis of hyperbilirubinemia in G6PD deficient neonates.....	52
14-	Morphological erythrocyte changes (Anisopoikilocytosis, bite cells) during acute hemolysis in G6PD-Deficient patient.....	63
15-	G6PD-Mediterranean C563T polymorphism (upper part).....	87
16-	G6PD silent (C1311T) polymorphism (upper part).....	88

17-	Correlation between 1311 silent polymorphism and TSB	94
18-	Correlation between Mediterranean 563 C/T mutation and total serum bilirubin.....	95
19-	Hemoglobin level and Mediterranean 563 C/T mutation	97
20-	Reticulocytic count and Mediterranean 563 C/T mutation	98
21-	Treatment and 1311 silent polymorphism.....	101
22-	Complications and 1311 silent polymorphism.....	103
23-	Complications and Mediterranean 563 C/T mutation.....	104
24-	Relationship between TSB and mild G6PD enzyme deficiency.....	107
25-	Relationship between TSB and moderate G6PD enzyme deficiency..	107
26-	Relationship between TSB and marked G6PD enzyme deficiency....	108

List of Abbreviations

AAP	:	American Academy of Pediatrics
ABE	:	Acute bilirubin encephalopathy
B/A ratio	:	Bilirubin/Albumin ratio
CNSHA	:	Chronic Non-spherocytic Hemolytic Anemia
CO	:	Carbon monoxide
DAT	:	Direct agglutinin test
ETCO	:	End tidal carbon monoxide
G6PD	:	Glucose-6-phosphate dehydrogenase
GA	:	Gestational Age
GST	:	Glutathione-s-transferase
Hct	:	Hematocrit
HGB	:	Hemoglobin
IVIg	:	Intravenous immunoglobulin
LED	:	Light Emitting Diodes
OATP2	:	Organic anion transport protein 2
PCR	:	Polymerase chain reaction
PCR-RFLP	:	Polymerase chain reaction- Restriction Fragment Length Polymorphism
snMP	:	Tin-Mesoporphyrin
TCB	:	Transcutaneous bilirubin
TSB	:	Total serum bilirubin
UGT	:	Uridine diphospho glucuronosyl transferase
WHO	:	World Health Organization

Aim of Work

The present study aimed to ascertain the presence of glucose -6-phosphate dehydrogenase (G6PD) enzyme deficiency in male newborns presented with neonatal hyperbilirubinemia, and to detect the presence or absence of G6PD Mediterranean mutation (563 C-T) and 1311 silent polymorphism. As well as, to delineates the magnitude of the problem of G6PD deficiency in Egyptian neonates, and emphasizes the role of early detection of G6PD deficiency by proper screening.