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Analysis of Neonatal Hyperbilirubinemia Caused by Variations in the G6PD Gene Sequence at RSIA Qurrata A'yun Samarinda

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Abstract. Hyperbilirubinemia is a common problem in newborns that is characterized by jaundice caused by high levels of bilirubin in the blood. One of the causes of hyperbilirubinemia is a deficiency in the enzyme glucose-6-phosphate dehydrogenase (G6PD), which is the most common metabolic disorder affecting over 400 million people worldwide. The goal of this study was to look at the G6PD gene variation in neonatal hyperbilirubinemia at the Qurrata A'yun Women's and Children's Hospital in Samarinda. This study employed an observational cross-sectional design. G6PD gene samples were obtained from 1–3 day old hyperbilirubin neonatal samples of 27 RSIA Qurrata A'yun Samarinda patients with bilirubin levels (> 10 mg/dl). Blood samples were examined using the G6PD humasis rapid test, which was obtained from 16 patients with G6PD deficiency and 11 normal patients. The G6PD gene amplification results were obtained in the 700 bp band, and the DNA coding for the G6PD gene was sequenced. G6PD gene mutation carriers were identified in 16 hyperbilirubinized neonatal samples. The most common gene mutation variants found in neonatal hyperbilirubin patients are Asahikawa (T 695 C), which causes amino acid changes (Leucine 232 Proline). In this study, 5 samples were obtained that indicated Sickle-Cell Anemia, demonstrating that hyperbilirubinized patients are inextricably linked to the anemia factor. This study found gene variants in the form of deletions, silent mutations, substitutions, and missense mutations, according to Asahikawa (T 695 C). In addition, one new mutation has been discovered, which was a change in the amino acid (Threonine 234 Glycine) or (C 701 G).

Keywords: Neonatal · G6PD · Hyperbilirubinemia, · Mutation

1 Introduction

Hyperbilirubinemia, often known as jaundice, is a medical condition in which the blood's bilirubin concentration rises. It can also be seen anatomically in neonates and results

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in yellow staining of the skin and eyes. Jaundice can result from hyperbilirubinemia, which is also a rise in blood bilirubin levels that can be brought on by a number of illnesses, including congenital defects [1]. The issue of hyperbilirubinemia frequently affects neonates. Jaundice is a sign of hyperbilirubinemia, which is defined by excessive levels of bilirubin in the blood. In infants, pathologically caused jaundice, also known as degrees three to five (>12 mg/dl) of jaundice, will emerge within the first 24 h of birth and is a complication of hyperbilirubinemia, namely kernicterus [2]. Blood type incompatibility, immaturity, infection, trauma, cephalic hematoma, some illnesses that result in erythrocyte abnormalities or erythrocyte biochemical deficiencies, and a lack of glucose-6-phosphate dehydrogenase (G6PD) are a few risk factors for hyperbilirubinemia in infants [3].

The most widespread metabolic condition, affecting more than 400 million people worldwide, is G6PD deficiency [4]. The most prevalent sex-related enzyme disease in humans is G6PD deficiency (x-linked). G6PD gene mutations are the primary cause of the basic metabolic abnormalities associated with G6PD deficiency. The pentosephosphate pathway's initial enzyme, G6PD, is responsible for converting glucose-6-phosphate to 6-phospho-gluconate during glycolysis. Nicotinamide Adenine Dinucleotide Phosphate (NADPH) is produced as a result of this transformation, and it lowers oxidized glutathione (GSSG) to reduced glutathione (GSH). The G6PD enzyme's role is to supply the NADPH required to rebuild GSH, which protects against hemolysis while maintaining the integrity of red blood cells. GSH works as a peroxide and H_2O_2 radical oxidant scavenger [5]. Genetic variation is any variation that happens in the nucleotide bases, genes, or chromosomes of an organism. Differences in the order of the nucleotide bases (adenine, thymine, guanine, and cytosine) that make up DNA in cells serve as an early marker of genetic variation (Harrison et al., 2014). Utilizing molecular markers, genetic variation can be analyzed. Indonesia has a prevalence of G6PD deficiency that ranges from 2.7 to 14.2% [6].

Elizabeth (2012) reported that 5.2% of 1802 neonates had a G6PD deficit. The majority of these infants were newborns, and little infants (those with birth weights under 2,500 g or gestational ages under 37 weeks) frequently had jaundice in the first several weeks of life. According to the 2012 Indonesian Demographic Health Survey (IDHS), there were 32 infant deaths for every 1000 live births. Asphyxia (37%) low birth weight (LBW) and prematurity (34%), sepsis (12%), hypothermia (7%), newborn jaundice (6%), postmaturity (3%), and congenital abnormalities (1% per 1000 live births) account for the majority of neonatal mortality in Indonesia [7].

The human G6PD gene, which has 13 exons, 12 introns, 2,269 bp of nucleotide mRNA, and 515 amino acids, is located on the X chromosome at position 28 on the q arm (Liliana, 2013). The enzyme's active form, known as G6PD, is made up of two or four identical subunits, each of which has a molecular weight of about 59 kDa. Mutations in the G6PD gene lead to impairments in protein function [8].

This gene's mutations result in G6PD deficiency. Since males only have one X chromosome, the presence or absence of the defective G6PD gene on that X chromosome determines whether a male has G6PD deficiency. Women can be normal homozygotes, G6PD deficient homozygotes, or G6PD deficient heterozygotes because they have two X chromosomes. The intermediate phenotype also refers to the heterozygous state [7].

2 Methodology

There are several tests available to identify G6PD deficiency. There are benefits and drawbacks to each test. Elizabeth (2012) claimed that there are numerous methods, including qualitative, quantitative, and molecular, that can be used to detect this.

An in vitro diagnostic test called a rapid diagnostic test is made for quick qualitative detection. The G6PD Humasis Rapid Diagnostic Test trademark is used for the actual G6PD test. The G6PD enzyme's activity is controlled by the G6PD humasis. The formazan technique is the foundation of this test. Blue dye is added to whole blood, and the reagent is then applied to the testing device. According to the calculation of the enzymatic reaction, the reaction will become colorless. When the sample's G6PD enzyme activity is normal, the test/test zone will turn red. Depending on the severity of the enzyme deficiency, the test zone will change from olive to blue-green in hue. When the control zone, which is the upper half of the test window, turns, the test is considered valid. A molecular technique**,** DNA analysis is unaffected by the hemolytic process thanks to PCR technology.

Polymerase Chain Reaction (PCR) is a method of multiplying certain DNA fragments exponentially. The DNA polymerase enzyme performs denaturation, annealing, and extension/elongation as part of the PCR process' basic repeated cycle process. A little amount of DNA template is used in the beginning of PCR, and millions of copies of the DNA are produced after numerous amplification cycles [9, 10]. PCR is used for the G6PD gene amplification process. The elements needed for PCR are. The target DNA strand is given a 5' end to the 3' end using a particular set of oligonucleotide primers, which amplifies the desired sequence. The G6PD gene was amplified using a PCR procedure that included 30 °C for 10 min.

Cycles of denaturation at 95 °C for 30 s, annealing at 55 °C for 30 s, and extension at 72 The length of the required DNA size as an amplification product determines the time. Using the reference gene bank NC 000023.11 (https://www.ncbi.nlm.nih.gov/), the G6PD gene was annotated, and a 700 bp G6PD primer was created using geneious software (https://www.geneious.com).

Purified DNA from PCR products is sent for the purpose of sequencing, which determines the pure DNA sequence. The National Center for Biotechnology Information (NCBI), National Institute for Health, USA, uses the Bioedit software Mega XI to analyze the rough data from the sequencing results. The processed data is then compared with the data in the gene bank using the Basic Local Alignment Search Tool (BLAST), which reveals the genetic relationship between the two species [12].

3 Result and Discussion

As many as 27 samples with newborn hyperbilirubinemia that were treated in the PBRT room at Qurrata A'yun Women's and Children's Hospital (WCH) Samarinda contained G6PD gene samples. A sample of venous blood up to 2–3 ml was drawn into an EDTA or N'Adactive tube using a 3 ml syringe, and the bilirubin level was then determined to be $>$ 10 mg/dl. The G6PD humasis quick test was used to analyze the blood sample, and the findings showed G6PD deficiency (16 samples) and normal (11 samples). The Favorgen Biotech Corp kit was also used to extract the DNA sample. If the OD260/280 ratio of a sample of DNA is 1.8 to 2.0, it is considered to be pure. The outcomes of the DNA extraction served as a template for 4_144_F(5'TTCAGCCCCATCTTAGCAGC3'), 4_833_R (5'GTCCCCAGCCACTTCTAACC3'). Results of G6PD gene amplification (700 bp). Additionally, samples of the PCR products were sent to PT. Indonesian Genetics for DNA sequencing to determine the location of the G6PD gene. Following this, data analysis was done on all the outcomes of the alignment process, BLAST (alignment of a sequence with other sequences that have been registered in the gene bank so that it can be seen which sequence has the highest percentage of similarity with the sequence).

Table 2 displays the results of the measurement of bilirubin in neonatal patients over the course of 1–3 days at Qurrata A'yun WCH Samarinda, followed by RDT G6PD testing and PCR for the G6PD gene.

3.1 Bilirubin and RDT G6PD Analysis

According to the analysis of bilirubin and RDT G6PD in newborns, 27 of the samples had bilirubin levels greater than 10 mg/dl. Neonatal jaundice, often known as neonatal jaundice, is a medical condition that frequently affects newborns. Bilirubin levels greater than 10 mg/dL are indicative of neonatal jaundice. Due to an overabundance of bilirubin, the disease known as jaundice causes the skin or other organs to discolor yellow. Jaundice affects about 60% of newborns who are healthy. Generally speaking, increased bilirubin levels are safe and don't need to be treated. However, certain occurrences are linked to many illnesses, including infections, endocrine, hepatic, and metabolic abnormalities as well as hemolytic disease [3]. Bilirubin levels greater than 20 mg/dL can cross.

Most newborns with G6PD deficiency experience early-onset jaundice, especially those who are little (birth weight 2,500 g or gestational age 37 weeks).

3.2 PCR G6Pd Dna

There were 27 samples of hyper bilirubin following RDT G6PD, 16 of which had deficiencies, and 11 of which had normal levels. Furthermore, PCR was performed on each sample for the G6PD gene. According to Fig. 1 and Table 4, the PCR findings revealed 16 positive samples with the appearance of a band of 700 bp and 10 negative samples (no PCR result bands occurred). The findings of the PCR test revealed a band of around 700 bp that matching gene mapping suggested might be a mutation [13].

Table 1. Neonatal hyperbilirubinemia at Qurrata A'yun WCH Samarinda was investigated using bilirubin, RDT G6PD, and PCR of the G6PD gene.

No	Samples	Bilirubin mg/dl		Rapid Test G6PD		PCR		Size (bp)	Seq
		Total	Direct	Deficiency	Normal	Positive	Negative		
1	B1	14.7	1.9		√	√		700	HBL1
2	B2	25.1	2.0	√		√		700	HBL2
3	B3	15.1	1.0	√		√		700	HBL3
4	B4	17.5	1.2	√		√		700	HBL4
5	B5	14.6	1.3	√		√		700	HBL5
6	B6	20.6	2.1		√	√		700	HBL6
7	B7	17.7	2.0		√	√		700	HBL7
8	B8	10.5	1.6		√		√	-	-
9	B9	22.3	2.3		√		√	-	-
10	B10	12.2	1.7	√		√		700	HBL8
11	B11	12.0	1.2	√		√		700	HBL9
12	B12	20.6	2.5		√		√	-	-
13	B13	10.3	1.2	√		√		700	HBL10
14	B14	21.5	2.1	√			√	-	-
No	**Sample**	**Bilirubin mg/dl**		**Rapid Test G6PD**		**PCR**		**Ukuran (bp)**	**Seq**
		Total	**Direct**	**Deficiency**	**Normal**	**Positive**	**Negative**		
1	B1	14.7	1.9		√	√		700	HBL1
2	B2	25.1	2.0	√		√		700	HBL2
3	B3	15.1	1.0	√		√		700	HBL3
4	B4	17.5	1.2	√		√		700	HBL4
5	B5	14.6	1.3	√		√		700	HBL5
6	B6	20.6	2.1		√	√		700	HBL6
7	B7	17.7	2.0		√	√		700	HBL7
8	B8	10.5	1.6		√		√	-	-
9	B9	22.3	2.3		√		√	-	-
10	B10	12.2	1.7	√		√		700	HBL8
11	B11	12.0	1.2	√		√		700	HBL9
12	B12	20.6	2.5		√		√	-	-
13	B13	10.3	1.2	√		√		700	HBL10
14	B14	21.5	2.1	√			√	-	-
15	B15	13.5	1.4		√	√		700	HBL11
16	B16	11.5	1.7	√			√	-	-
17	B17	11.8	1.5	√		√		700	HBL12
18	B18	13.5	1.2	√		√		700	HBL13
19	B19	21.3	2.7	√		√		700	HBL14
20	B20	19,4	2.5		√	√		700	HBL15
21	B21	23.0	2.1	√		√		700	HBL16

(*continued*)

Table 1. (*continued*)

No	Sample	Bilirubin mg/dl		Rapid Test G6PD		PCR		Ukuran (bp)	Seq
		Total	Direct	Deficiency	Normal	Positive	Negative		
22	B22	16.3	2.0	√		√		700	HBL17
23	B23	14.1	1.5	√			√	-	-
24	B24	10.7	1.3		√		√	-	-
25	B25	15.5	2.1		√		√	-	-
26	B26	11.8	1.8		√		√	-	-
27	B27	24.7	2.3	√			√	-	-
TOTAL				**16**	**11**	**17**	**10**		**17**

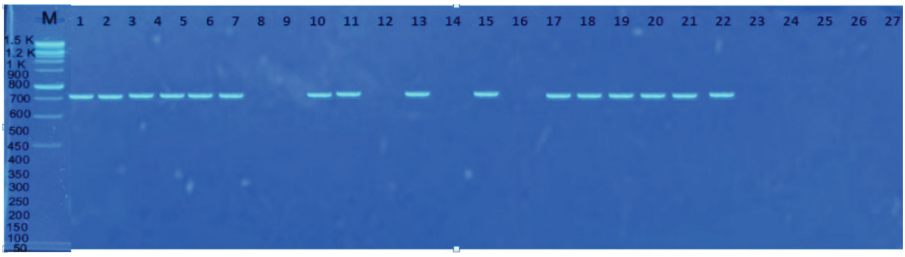

Fig. 1. Neonatal hyperbilirubin PCR results at Qurrata A'yun WCH Samarinda. M (Marker)1.5Kb, Normal RDT with Positive PCR (1, 6, 7, 15, and 20), Deficiency RDT with Positive PCR (2, 3, 4, 5, 10, 11, 13, 17, 18, 19, and 21), and Normal RDT with Negative PCR (8, 9, 12, 24, 25, and 26). (14,16,23 and 27).

3.3 DNA Sequencing of G6PD Gene

The MEGA XI program was used to build consensus on the forward and reverse sequences of the G6PD gene. The G6PD gene's DNA sequencing findings were examined, compared to the information in the Gen Bank Basic Local Alignment Search Tool (BLAST) at NCBI, and results were obtained. The information showing that strain 1256 and strain 1256 have 99% identity in the G6PD gene's DNA sequence. Following the mapping of the G6PD gene's DNA sequencing results to reference genes, the data was examined as follows: (Table 1).

3.4 Gene Reference Mapping

Table 2 showed the findings of the reference gene mapping in neonatal hyperbilirubin**.**

According to Table 3, there are substitutions at positions 14 and 643, 692, 696, 697, 701, 702, 703, and 704. There are also a number of deletions at these positions. Sequences of 615, 643, 645, and 700 silent mutations. Additionally, missense mutations exist at positions 695, 696, 701, and 703. Table 3 displays the amino acids produced based on modifications to the nucleotide bases.

Table 2. The mapping of the G6PD gene nucleotide bases on newborn hyperbilirubin at Qurrata A'yun (WCH) Samarinda.

Nucleotide sequence	14	615	643	645	692	695	696	697	700	701	702	703	704	Information			
G6PD	A	C	T	G	G	T	G	G	A	C	C	G	A	mutation		Impact	
Sample Code														S	D	SM	MM
B1	A	C	C	T	G	C	T	G	G	G	C	-	-	6	2	5	1
B2	A	T	-	G	G	T	-	-	G	G	C	A	A	4	3	3	1
B3	A	C	T	G	-	T	T	G	G	G	C	A	A	4	3	3	1
B4	A	T	T	G	G	C	T	G	G	G	C	-	-	5	2	4	1
B5	T	C	T	G	G	T	T	G	G	-	C	-	-	2	3	1	1
B6	T	C	T	G	-	C	T	G	G	G	C	A	-	6	2	5	1
B7	T	C	T	G	G	C	T	G	G	G	C	-	-	5	2	4	1
B11	T	C	T	G	G	C	T	G	G	G	C	-	-	5	2	4	1
B13	T	C	T	G	G	C	T	G	G	G	C	-	-	5	2	4	1
B15	A	C	T	G	-	C	T	G	G	G	C	-	-	4	2	3	1
B17	A	C	T	G	G	C	T	G	G	G	-	-	-	4	4	3	1
B18	A	C	T	G	-	C	T	G	G	G	C	-	-	4	3	3	1
B19	A	C	T	G	-	C	T	G	G	G	C	-	-	4	3	3	1
B21	A	C	T	G	-	C	T	G	G	G	C	-	-	4	3	3	1
B22	A	C	T	G	-	C	T	G	G	G	-	-	-	4	4	3	1

The outcomes of the above-mapped data's sequencing are next examined for nucleotide bases and amino acids as follows: Table 4 shows an analysis of the mutation data from the G6PD gene sequencing variation of nucleotide bases and amino acids.

There are seven different types of G6PD mutants in Surabaya, according to Kurniawan's (2014) research; in this study, all of them were categorized as point mutations, including a) 4 cases of G6PD Vanua Lav, b) 1 case of G6PD Viangchan, and c) 5 cases of G6PD Chatham. d) 2 G6PD Union cases, e) 7 G6PD Canton cases, f) 6 G6PD Kaiping cases, and g) 2 G6PD Silent cases. According to findings from earlier research and the current study, more than 90 mutations have been identified at the DNA level, the majority of which take the form of point mutations while others take the form of base pair deletions [6]. It's intriguing that the primary mutation is a CpG dinucleotide shift from C to T that is supposed to stand for [13].

Table 3. Deletions of G6PD Genes (mutations)

No	Mutation	Nucleotides in DNA	Acid Amino	Information
1	Deletion	643 T → - 692 G → - 695 T → - 696 G → - 697 G → - 701 C → - 702 C → - 703 G → - 704 G → -	214 Phenylalanine → Phenylalanine 231 Trypyophan → Trypyophan 232 Leucine → Leucine 232 Leucine → Leucine 233 Glysine → Glysine 234 Threonine → Glycine 234 Threonine → Threonine 234 Threonine → Glycine 235 Arginine → Arginine	Base nucleotide missing
2	Silent mutation	615 C → T 643 T → C 645 G → T 700 A → G	205 Leucine → Leucine 214 Phenylalanine → Phenylalanine 215 Leucine → Leucine 233 Glysine → Glysine	has no effect on amino acids (Substitution)
3	Substitution	14 A → T	5 Histidine → Leucine	Sickle-Cell Anemia
4	Missense mutation	695 T → C 696 G → T 701 C → G 703 G → A	232 leucine → Proline 232 leucine → Proline 234 Threonine → Glycine 234 Threonine → Glycine	The amino acid was changed (Substitution)
5	Asahikawa	695 T → C	232 Leucine → Proline	

In this investigation, DNA samples of neonatal hyperbilirubin G6PD with five mutation variants, such as: deletion, silent mutation, substitution, missense mutation, and Asahikawa (T 695 C) amino acid were collected at RSIA Qurrata A'yun Samarinda, East Kalimantan (Leucine 232 Proline). They are considered to be point mutations when looking at variants in terms of DNA level classification. The G6PD gene region has 5 different mutation variants, according to the findings of the genome-based mapping that was done.

This study's mutation was not found in earlier research done at Qurrata A'yun WCH Samarinda, where the key variant found was the amino acid Asahikawa (T 695 C) (Leucine 232 Proline). Twelve of the 17 gene mapping samples, which were compatible with the DNA-level characterization of the G6PD variation, showed changes in the sequence of nucleotide bases and amino acids. The substitution of the amino acid (A 14 T) (Histidine 5 Leucine) from one of the five samples on the map indicated sickle-cell anemia [14–16]. Deletion mutations, Silent mutations, Substitutions, and Missense mutations produce outcomes that are comparable to those of earlier research.

Table 4. Results of G6PD gene mutations and variations in newborn hyperbilirubin at Qurrata A'yun WCH Samarinda.

No	**Alternate Name**	**Nucleotides in DNA**	**Acid Amino**	**Information**
1	Deletion	643 T → - 692 G → - 695 T → - 696 G → - 697 G → - 701 C → - 702 C → - 703 G → - 704 G → -	214 Phenylalanine → Phenylalanine 231 Trypyophan → Trypyophan 232 Leucine → Leucine 232 Leucine → Leucine 233 Glysine → Glysine 234 Threonine → Glycine 234 Threonine → Threonine 234 Threonine → Glycine 235 Arginine → Arginine	Base nucleotide missing
2	Silent mutation	615 C → T 643 T → C 645 G → T 700 A → G	205 Leucine → Leucine 214 Phenylalanine → Phenylalanine 215 Leucine → Leucine 233 Glysine → Glysine	has no effect on amino acids (Substitution)
3	Substitution	14 A → T	5 Histidine → Leucine	Sickle-Cell Anemia
4	Missense mutation	695 T → C 696 G → T 701 C → G 703 G → A	232 leucine → Proline 232 leucine → Proline 234 Threonine → Glycine 234 Threonine → Glycine	The amino acid was changed (Substitution)
5	Asahikawa	695 T → C	232 Leucine → Proline	

4 Conclusion

According to the research, the neonatal hyperbilirubin G6PD DNA sample at RSIA Qurrata A'yun Samarinda, East Kalimantan, has six variants: deletion, silent mutation, substitution, missense mutation, and asahikawa (T 695 C) amino acid (Leucine 232 proline). Five mutations were discovered in the G6PD gene area based on the mapping results obtained utilizing the genome.

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Author's Contribution. Conceptualization: PS, AR, SD and MDK; Methodology: PS, AR, Formal analysis and validation: PS, MDK, SD; Preparation: AR, Writing: MDK.

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Dear Prince Saputra

Congratulation!

We are pleased to inform you that your abstract with entitled "**Analysis of Neonatal hyperbilirubinemia caused by Variations in the G6PD Gene Sequence at RSI Qurrata'ayun Samarinda**" have been successfully accepted in the 1st Lawang Sewu International Symposium on Health Sciences 2022 organized by Universitas MuhammadiyahSemarang, Indonesia. Further information will be given through email. Should you have any other inquiries related to this confrence, please do not hesitate to contact us.

Regards,

Ns. Satriya Pranata, M.Kep.,PhD
Chairman of The 1st Lawang Sewu
International Symposium on Health Sciences