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CANTON AND MEDITERRANEAN MUTATIONS AMONG CHINESE AND MALAY NEWBORNS WITH G6PD ABNORMALITIES IN UKMMC

By

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DECLARATION

I hereby declare that this thesis is based on my original work and has not been submitted previously or currently for any other degree at UiTM or any other institutions.

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ABSTRACT

Canton and Mediterranean mutations among Chinese and Malay newborns with G6PD abnormalities in UKMMC

Glucose-6-Phosphate Dehydrogenase (G6PD) is an enzyme that helps in the protection of red blood cells against oxidative stress. G6PD deficiency, which is an X-linked genetic disorder, is the commonest enzyme defect in human. It will cause erythrocytes to breakdown prematurely and lead to a hemolytic condition. There are several classes of genetic variants for G6PD deficiency. The variants are classified according to severity of deficiency. Determination of the severity will help in monitoring and prevention from occurrence of complication such as kernicterus which can lead to death. In order to identify the type of variant, molecular technique is the method of choice. However, prior to the implementation of any molecular method, data on the percentage of common G6PD mutations for a specific population within a geographic area is required. Therefore, the study was conducted to detect the presence of Canton and Mediterranean mutations among Chinese and Malay newborns with abnormal G6PD in UKM Medical Centre. Cord blood samples from newborns with abnormal G6PD (80 Malay and 23 Chinese) were selected based on their FST and G6PD enzyme level assay results. The detection of mutations was performed using 7500 Fast Real-Time Polymerase Chain Reaction. The results showed that the Mediterranean mutation was present in twelve of the Malay newborns while the Canton mutation was detected in six of the Chinese newborns. Out of the twelve Malay newborns, ten were detected as homozygote, while two newborns were detected as heterozygote. Meanwhile, out of six Chinese newborns, three were heterozygote and three were homozygote. As for the remaining sixty-eight Malays newborns and seventeen Chinese newborns, they were detected as wild-type. However, beside these two mutations, there are other common G6PD mutations. Thus, there is a high possibility that the undetected newborns may have one of the other common mutations which were not screened in the present study. In conclusion, the Canton and Mediterranean mutations were detected among Chinese and Malay newborns with abnormal G6PD status in UKMMC with prevalence of 26.09% and 15% respectively.

Keywords: Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency; G6PD Canton; G6PD Mediterranean; 7500 Fast Real-Time Polymerase Chain Reaction

CHAPTER 1

INTRODUCTION

1.1 Background of study

Glucose-6-Phosphate Dehydrogenase (G6PD) is an enzyme that can be found in human erythrocytes. This enzyme is an enzyme that helps in the protection of red blood cells against oxidative stress. Defect of this enzyme will cause the erythrocytes to hemolyse prematurely. Hence, it results in a hemolytic condition. The hemolysis of the erythrocytes can increase the level of bilirubin and thus will lead to several complications such as neonatal jaundice, hemolytic anemia, congenital non-spherocytic hemolytic anemia and favism.

G6PD deficiency is the most common enzyme defect in humans. It is an inherited condition and is known as an X-linked genetic disorder. According to Nadarajan *et al.* (2011), this genetic disorder is most common in people with African, Asian, Middle Eastern and Mediterranean or Middle Eastern descent. This deficiency was estimated to affect about 400 million individuals worldwide (Ainoon *et al.*, 2004). According to Ainoon *et al.* (2003), the prevalence of G6PD deficiency in Malaysia is 7.17%. Studies have also revealed that the prevalence of G6PD deficiency among male according to ethnicity were 4.6%, 6.0%, 1.3% while among females were 1.4%, 1.6% and 0.49% in Malays, Chinese and Indians, respectively (Ainoon *et al.*, 2004). Based on these studies, it was shown that G6PD deficiency was more prevalent in males and among the Malays and Chinese and less common in Indians.

G6PD deficiency results from the synthesis of structurally abnormal enzyme variants of the G6PD gene. Previous studies have found various types of mutation within the gene that can cause G6PD deficiency. Different types of mutations will create different variants and different classes of enzyme activity