국내 다문화 소아청소년의 Glucose-6 Phosphate Dehydrogenase 결핍 유병률

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The Prevalence of Glucose-6 Phosphate Dehydrogenase Deficiency among Multi-Cultural Youths in Korea

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Dear Editor

Hereditary hemolytic anemia is genetically and phenotypically diverse, with α - and β - hemoglobino-pathies, glucose-6-phosphate dehydrogenase (G6PD) deficiency, and hereditary spherocytosis being the most common forms worldwide [1]. A G6PD deficiency commonly results from an adaptation to malaria infections and is most common in the tropical and subtropical regions of the Eastern Hemisphere (Africa, Europe, and Asia) [1]. Its prevalence are 8.5%, 5.2%, 3.8%, and 7.2% in Africa, the USA and Asia, Europe, and the Middle East, respectively [1]. A previous study failed to detect any known G6PD

deficiency variants in the Korean population [2].

G6PD-deficient erythrocytes can be destroyed more easily through oxidative stress than normal erythrocytes, owing to the decreased nicotinamide adenine dinucleotide phosphate levels. In patients with a G6PD deficiency, hemolysis occurs when the red blood cells encounter oxidative stress due to drugs or fava bean ingestion, and infections [3]. Patients with unexplained occurrences of hemolytic anemia, jaundice, or dark urine should be evaluated for a G6PD deficiency. Red blood cell hemolysis occurs in patients receiving drugs that cause oxidative stress after being transfused blood from patients with a

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G6PD deficiency [3]. Therefore, a G6PD deficiency should be diagnosed to help ensure the patients' and transfusion recipients' well-being.

Nowadays, the number of immigrants is increasing continuously in Korea, resulting in wider ethnic diversity [4]. The incidence of G6PD deficiency was investigated among volunteers of multi-cultural youth to determine the G6PD deficiency screening requirements. This was a prospective and observational multi-center study conducted from September 2015 to August 2017. A total of 68 multi-cultural youth (health volunteers and patients aged <30 years), who self-identified their ethnicity at seven hospitals in Korea, were recruited and enrolled in this study. The peripheral blood was collected into EDTA-containing tubes, and the CBC was obtained using an automated cell counter (Sysmex XN-3000, Sysmex Corporation, Kobe, Japan). Hemoglobin (Hb), mean

corpuscular volume (MCV), and red cell distribution width-coefficient of variation (RDW-CV) were recorded. G6PD variants were detected using a DiaPlexC G6PD Genotyping Kit (SolGent, Daejeon, Korea), which allows the screening of the seven variants of the G6PD gene using an allele-specific multiplex PCR. Multiplex PCR can detect multiple specific target genes in a single PCR. The seven variants were Vanua Lava (383 T>C), Mahidol (487 G>A), Mediterranean (563 C>T), Coimbra (592 C>T), Viangchan (871 G>A), Union (1360 C>T), Canton (1376 G>T), and Kalping (1388 G>A). After extracting the genomic DNA from 50 blood samples using a QuickGene DNA Whole Blood Kit S (KURABO, Osaka, Japan), a PCR mixture was prepared using the DiaPlexC kit, and 200 ng of the total genomic DNA was added to 25 µL of the master mixture. PCR was performed based on the cy-

Table 1. Baseline characteristics of the study groups

Parameters			Results, median (range)
Age (months)			7 (0~341)
Sex	Female		32
	Male		36
RBC indices	Hemoglobin (g/dL)		13.0 (6.2~19.7)
	MCV (fL)		86.36 (49.5~128.2)
	RDW-CV (%)		15.1 (11.3~21.4)
Nationality of one or more parents (Except Korea)	Southeast Asia	Vietnamese	38
		Philippines	7
		Cambodia	3
		Thailand	1
		Laos	1
	Northeast Asia	China	10
		Other	2
	South Asia		1
	Europe, America, Africa		5

Abbreviations: MCV, Mean Corpuscular Volume; RDW-CV, Red Cell Distribution Width-coefficient of variation.

clinging conditions recommended by the manufacturer. The PCR products were then resolved by electrophoresis on a 2% agarose gel, stained with RedSafe (iNtRON Biotechnology, Seongnam, Korea), and visualized under ultraviolet illumination. Each PCR was normalized using an internal control. Table 1 lists the characteristics of the study groups. Owing to the enrollment of 38 infants, the median age was 7 months old; 55.9% of the subjects had one or more parents from Vietnam. In this study, the incidence of a G6PD deficiency was found to be 0%.

The prevalence of G6PD deficiency is high in malaria-endemic countries. Using the assessment methods and regions of the study, the reported prevalence of G6PD deficiency ranges from 1.6~9.0% in China to 3.2~29.6% in Myanmar [1]. According to a recent report of the Ministry of Justice's Statistical Yearbook of Foreign Policy on Immigration, there are approximately 719,282 immigrants from China, Vietnam, Thailand, and Myanmar in Korea [4]. Based on the numerical calculation, with the known prevalence and residents in Korea, the number of potential G6PD deficiency carriers among multicultural youth was estimated to be at least 20,000. Therefore, a G6PD deficiency might increase in the near future.

A G6PD deficiency has an X-linked recessive mode of inheritance, and the mutation decreases the enzyme stability [1]. The son can acquire the disease if the mother is an asymptomatic carrier. The exact prevalence of G6PD deficiency in immigrants should be evaluated to establish appropriate public health policies depending on whether a G6PD deficiency could be a major cause of hereditary hemolytic anemia in Korea. Furthermore, G6PD deficient blood

can affect transfusion recipients suffering from hemolytic anemia. According to the 2012 WHO Guidelines for Assessing Donor Suitability for Blood Donation, patients with a G6PD deficiency should avoid lifetime blood donations if they have a history of hemolysis, and in areas with a high prevalence, screening tests for a G6PD deficiency are recommended [5]. Although the incidence of G6PD deficiency is increasing continuously, the total number of patients diagnosed is limited nationwide; therefore, a national public health policy aimed at preventing G6PD-deficient patients from donating blood should be implemented.

In this study, no known G6PD deficiency variants among multi-cultural youths were detected due to the small study population. To ensure the timely diagnosis and treatment of G6PD deficiency and prepare for the need for the screening of foreign donors in the future, it is necessary to identify the actual prevalence of G6PD deficiency in a large number of migrants from Southeast Asia.

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