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Molecular characterization of glucose-6-phosphate dehydrogenase deficiency specific variants in Amhara region, Ethiopia

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Background: Glucose 6-phosphate dehydrogenase deficiency (G6PDd) is an X-linked hereditary genetic defect, affecting an estimated 400 million people worldwide. Severe clinical manifestations associated with G6PDd (e.g., chronic hemolytic anemia) depends on the type of G6PD molecular variants and exposure to hemolytic triggers (e.g., antimalarial like Primaquine). However, a scarce study on G6PDd renders the use of Primaquine for effective therapeutic treatment of malaria.

Objectives: To determine the availability and characterization of selected molecular variants of G6PDd specific genes among selected populations in malaria endemic area of Amhara region, Ethiopia.

Methods: Using a cross sectional study design, a total of 156 dried blood samples were randomly selected from 360 stored samples of national malaria indicator survey of 2011 starting from July 30/2014 to January 30/2015. Polymerase chain reaction and restricted fragment length polymorphism technique was applied to characterize G6PDd variants as G6PD*A, G6PD*A- and/or G6PD*Mediterranean. Binary logistic regression was applied to see association (P<0.05 is significant) among different parameters.

Result: Of 156 studied dried blood spot samples, 10 (6.4%) had G6PD genotype available. G6PD*A (100%) was the only genotype characterized, while neither G6PD*A- nor G6PD*Mediterranean genotypes were detected. There was no statistical significant difference between G6PDd and other socio demographic and risk related variables (P>0.05).

Conclusion: G6PD*A variant was the only G6PDd genotype detected in this study. G6PD*A variant has almost (90%) the same enzymatic activities with the wild type. Therefore, this result supports the safe use of primaquine, especially the single low dose for transmission interruption of Plasmodium falciparum gametocyte and radical cure of *Plasmodium vivax*, as a part of malaria elimination toolkit, among selected populations in malaria endemic areas of Amhara region.

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