Comparison of fluorescent spot test, decolorization test and quantitative enzyme assay in detection of G6PD enzyme deficiency

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Abstract

Background and Objective: The national screening program for G6PD enzyme deficiency is not able to detect all affected neonates. This study was done to compare the fluorescent spot test (FST), decolorization test, and quantitative enzyme assay (QEA) for detecting G6PD enzyme deficiency in neonates.

Methods: In this descriptive study, cord blood samples of 365 neonates were collected. Decolorization test, QEA and DNA test was done for each sample. All of the neonates were tested by FST as a part of national screening program on heel-prick blood sample collected on day 3–5 after birth. QEA was considered as the gold standard. According to QEA test results, neonates with <20% and 20–60% of mean normal enzyme activity were considered as total deficient and partial deficient, respectively.

Results: Fluorescent spot test detected 13 male neonates with G6PD enzyme deficiency while decolorization test identified 18 male and 1 female neonates. Using QEA, 19 of male and 28 of female neonates with G6PD enzyme deficiency (26 cases with partial and 2 cases with total deficiency) were diagnosed. DNA analysis detected 34 female case as heterozygote and 14 male neonates as hemizygote for the disease.

Conclusion: Fluorescent spot test do not have required sensitivity for screening of neonates with G6PD enzyme deficiency. QEA test is recommended to replace the fluorescent spot test in national screening program.

Keywords: G6PD enzyme deficiency, fluorescent spot test, decolorization test, quantitative enzyme assay, DNA test

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