GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Screening and prevention of neonatal glucose 6-phosphate dehydrogenase deficiency in Guangzhou, China

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Genet. Mol. Res. 13 (2): 4272-4279 (2014)
Received May 21, 2013
Accepted October 4, 2013
Published June 9, 2014
DOI http://dx.doi.org/10.4238/2014.June.9.13

ABSTRACT. We aimed to summarize the results of screening protocol and prevention of neonatal glucose 6-phosphate dehydrogenase (G6PD) deficiency during a 22-year-long period to provide a basis of reference for the screening of this disease. About 1,705,569 newborn subjects in Guangzhou City were screened for this deficiency. Specimens were collected according to the conventional method of specimen acquisition for "newborn dried bloodspot screening", preserved, and inspected. The specimens were studied with fluorescent spot test and quantitative fluorescence assay. Diagnosis was performed using the modified NBTG6PD/6PGD ratio method. Bloodspot filter paper specimens were sent to the laboratory within 24 h via EMS Express, and the G6PD test was performed on the same day. The G6PD deficiency-positive rate was 4.2% in the samples screened using the fluorescent spot test, while it was 5% in case of the quantitative fluorescence assay. Neonatal screening for G6PD deficiency for 11,437 cases (6117 boys and 5320 girls) showed positive results in 481 cases. About 420 cases (318 boys and 102 girls) of G6PD deficiency were confirmed with the modified

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Duchenne NBT ratio method. The total detection rate was 3.7:5.2% for boys and 1.9% for girls. Quantitative fluorescence assay improved the sensitivity and detection rate. Accelerating the speed of sample delivery by using Internet network systems and ensuring online availability of screening results can aid the screening and diagnosis of this deficiency within 1 week of birth.

Key words: Glucose 6-phosphate dehydrogenase; Deficiency; Neonatal screening

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