Population-based Carrier Screening and Prenatal Diagnosis of Fragile X Syndrome in East Asian Populations

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Abstract

Identification of carriers of fragile X syndrome (FXS) with the subsequent prenatal diagnosis, and knowledge of FXS-associated genetic profiles are essential for intervention in specific populations. We report the results of carrier screening of 39,458 East Asian adult women and prenatal diagnosis from 87 FXS carriers. The prevalence of FXS carriers and incidence of full mutation fetuses in carrier pregnancies were found to be 1/556 and 11.0%, respectively. We confirmed the validity of the current threshold of CGG repeats for FMR1 categorization; the integral risks of full mutation expansion were approximately 6.0%, 43.8%, and 100% for premutation alleles with 55-74, 75-89, and [?]90 CGG repeats, respectively. The protective effect of AGG interruption in East Asian populations was validated, which is important in protecting premutation alleles with 75-89 CGG repeats from full mutation expansion. Lastly, family history was shown not an effective indicator for FXS carrier screening in East Asian populations and population-based screening was more cost-effective. This study provides an insight into the largest carrier screening and prenatal diagnosis for FXS in East Asian populations to date. The FXS-associated genetic profiles of East Asian populations are delineated and population-based carrier screening is shown to be promising for FXS intervention.

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