Genetic Background of Long QT Syndrome in Infants, Children, and Adolescents
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Introduction: Little data are available for the prevalence of LQT1, LQT2, and LQT3 genotypes among these three mutations in pediatric population. A school-based ECG screening program in Japan screened children with definite LQTS without past history of symptoms. We aimed to determine the characteristics of genetic background in childhood LQTS in Japan. Methods: The study population included 176 family members from 84 probands (0-17 years; median, 9.5 years; M:F=46:38) who were referred to our center from 1993-2010. Genomic DNA was isolated from blood and screening for LQT1-LQT12, except for LQT4&11. Results: Genotypes were identified in 51 of 84 probands and in 87 of 176 family members. LQT1 was found in 42 family members (from 24 probands), LQT2 in 22 (16), LQT3 in 18 (9), and others in 12 (6). The prevalence of LQT3 in probands (9/46) and in family members (18/80) in the present study was significantly higher (p=0.006 and p=0.002, respectively) than the data for the adult population (11/192 in probands and 82/812 in family members by Sauer AJ, et al). Of 84 probands, the screened subjects showed a higher rate of genotypic determination (30/38) than symptomatic subjects (15/35, p= 0.002). Conclusions: A high prevalence of the LQT3 genotype in the pediatric population suggests progress in the medical management of these patients during infancy and childhood. School-based ECG screening and genetic testing may help prevention of LQTS-related symptoms in Japan. Keywords: Long QT syndrome, genotype, school-based screening