Clinical Genetics

AB061. Prevalence of 22q11.2 deletion syndrome in patients with congenital heart diseases in North-eastern Thailand

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Background: 22q11.2 deletion syndrome is the most common microdeletion syndrome that leads to multisystem involvement especially cardiovascular malformations. Frequencies of this syndrome in patients with cardiovascular malformations are different in various ethnic groups. The objective of this study was to determine the prevalence of this syndrome in North-eastern Thai population with congenital heart diseases.

Methods: Medical records of 359 patients with congenital heart diseases at Srinagarind Hospital in 2016 were retrospective reviewed. Eighty-three patients were evaluated for 22q11.2 deletion syndrome by fluorescence *in situ* hybridization (FISH) analysis.

Results: Twenty-four of 83 (28.9%) patients with cardiovascular malformations were found with 22q11.2 deletion syndrome. Nineteen of 56 (33.9%) patients with conotruncal heart defects (95% CI: 21.8–47.8) and 5 of 27 (18.5%) patients with non-conotruncal heart defects were found to have the deletion (95% CI: 6.3–38.1). The 22q11 deletion were found in 14 of 24 (58.3%) patients with tetralogy of Fallot (TOF) (95% CI: 36.6–77.9), 3 in 12 (25%) patients with atrial septal defect (ASD) (95% CI: 5.5–57.2), 1 in 7 (14.3%) patients with pulmonary stenosis (PS) (95% CI: 0.4–57.9), 1 in 4 (25%) patients with transposition of great arteries (TGA) (95% CI: 0.6–80.6) and 1 in 4 (25%) patients with ventricular septal defect (VSD) (95% CI: 0.6–80.6). One of two patients with truncus arteriosus and one out of two patients with double outlet of right ventricle and pulmonary atresia with VSD were positive for the deletion.

Conclusions: The prevalence of 22q11.2 deletion syndrome is common among patients with congenital heart disease in North-eastern Thailand. Half of the patients with TOF were found associated with this syndrome. The 22q11.2 deletion syndrome not only had association with conotruncal heart diseases, but also non-conotruncal or acyanotic heart diseases. Further study with a larger scaled population is recommended to determine the real advantage of screening for 22q11.2 deletion syndrome in congenital heart diseases.

Keywords: 2q11.2 deletion syndrome; congenital heart disease; tetralogy of Fallot (TOF); velocardiofacial syndrome

doi: 10.21037/atm.2017.s061

Cite this abstract as: Srichaisawat P, Wichajarn K, Chaikitpinyo A, Panamonta M, Kampan J. Prevalence of 22q11.2 deletion syndrome in patients with congenital heart diseases in North-eastern Thailand. Ann Transl Med 2017;5(Suppl 2):AB061. doi: 10.21037/atm.2017.s061

