Tran Quynh Nhu Nguyen et al., Brain Disord Ther 2016, 5:3(Suppl)
http://dx.doi.org/10.4172/2168-975X.C1.011

Congenital heart disorder is a high risk of developmental disabilities

Tran Quynh Nhu Nguyen[1,2], Makiko Saitoh[1], Huu Tung Trinh[2], Nguyen Minh Thien Doan[2], and Masashi Mizuguchi[1]
[1]University of Tokyo, Japan
[2]Children's Hospital, Vietnam

Ellis-van Creveld syndrome (EvC) is a ciliopathy with cardiac anomalies, disproportionate short stature, polydactyly, dystrophic nails and oral defects. Approximately 60% of EvC patients have severe congenital heart disorders (CHDs), of which more than half are atrio-ventricular septal defect (AVSD) and common atrium. Neuropsychological development of EvC is generally normal. However, CHD was confirmed as a high risk of developmental disabilities (DDs). Here we presented a typical EvC case with DDs. She is a 2.5-year-old female, the first child of consanguineous couple. Here height was 62.0 cm (under the 2nd percentile), post-axial polydactyly of hands, nail dystrophy, and AVSD with pulmonary stenosis. Her SPO2 was 70-80% from birth. We detected two novel compound heterozygous variants (p. E177X, p.R826X) in this patient. Her mental development was normal up to the age of 2 years old. From 2 to 2 ½ years-old, she reveals DDs, which was evaluate by ASQ-3 score. The head circumference is 47.0 cm. MRI showed no defects. We suggested that hypoxia caused DDs in this patient. DDs in children with CHD are common and should be alert. The feeding difficulty, hypoxia, medical comorbidities, genetic abnormalities, and more complex treatment increased risk for DD. Thus, longitudinal follow-up throughout childhood and into adulthood is necessary for children with CHD because exposure to risk and prevalence of DD change over time. Primary and special care for children with CHD is critical to minimize DDs. (233 words)

Biography

Nguyen Tran Quynh Nhu graduated from University of Medicine and Pharmacy, Ho Chi Minh city, Vietnam in 2007. Since 2008, she has worked as cardiologist in Children's Hospital 2, Ho Chi Minh city, Vietnam. She has completed master course from School of Medicine, The University of Tokyo, Japan in March 2015 and now continues Ph.D course at the same above university.

qnhu13@gmail.com

Notes: