Current Pediatric Research

Sex differences in congenital heart disease in Japanese Down Syndrome

Takako Takano

Japan Pediatric Society, Japan

Abstract

Down syndrome (DS) is one of the most common chromosomal abnormalities, which is associated with various unique physical traits and developmental delay. As the mortality rate of people with DS has declined, life expectancy at birth of people with DS was increased between recent 50 years. Life expectancy for the time period of 1953 to 2000 was 58.6 year, compared with 16.2 years for the period of 1948 to 1957 among those registered in a DS database in Western Australia. In Japan, the life expectancy for 1,052 DS who were born between 1966 and 1975 increased to 48.9 years. In Western Australia, in contrast to female longevity in the general population, males with DS were found to have significantly longer life expectancies, compared with their female counterparts. We investigated sex differences in congenital heart disease (CHD), which may be related to mortality using data on 1310 people with DS (626 females and 684 males) in Japan from five hospitals' medical records and questionnaires completed by parents of people with DS. The CHD rate was significantly higher for females (354, 57%) than for males (338, 49%; p=0.010). Significantly more females (199, 32%) than males (175, 26%) underwent surgery for CHD (p=0.018). Higher prevalence and greater severity of CHD in females may contribute to poor prognoses. We found that the most common cardiac anomalies (main lesions) were VSD, ASD, AVSD, PDA and TOF, which together accounted for 95.5% of all CHD cases. Among people with DS born before 1979, in the1980s or 1990s, we found that significantly more female than male subjects had CHD, but we found no significant sex difference for subjects born in 2000 or later. Many factors seem to have contributed to this change, including the improvement of diagnostic techniques, such as echocardiographic examination, and improvements in heart surgery. This shift may be attributed to a great increase in the diagnosis of less severe CHD for both sexes. Improvements in quality of life of individuals with DS have resulted from prognosis in medical care, identification and treatment of CHD in perinatal and postnatal period.

Biography

Takako Takano is a professor at Department of Child Health, Tokyo Kasei University and Graduate School of Human Life Sciences, Tokyo Kasei University, Tokyo, Japan. She is also an Adjunct lecturer at the Department of Human Genetics, Graduate School of Medicine, the University of Tokyo. Takano grew up in Tokyo and graduated from Faculty of Medicine, the University of Tokyo, where she received a PhD degree in 1985. She has worked as a Pediatrician. After training of cytogenetics, especially chromosome research at National Institute of Genetics, her main field of research is cytogenetics and clinical genetics. During her 13-year career at Teikyo University School of Medicine, she studied FISH (fluorescence in situ hybridization) and applied this technique for clinical genetics. Takano continues clinical examinations and genetic counseling for disabled children with genetic diseases or congenital abnormalities. She is a member of the Japan Society of Human Genetics, the Japan Society of Pediatric Genetics, Japan Pediatric Society and the Japanese Society of Child Health.

Publication

1. Glasson EJ, Sullivan SG, Hussain R, et al. The changing survival profile of people with Down's syndrome: implications for genetic counselling.
2. Masaki M, Higurashi M, Iijima K, et al. Mortality and survival for Down syndrome in Japan.
3. Hijii T, Fukushige J, Igarashi H, et al. Life expectancy and social adaptation in individuals with Down syndrome with and without surgery for congenital heart disease.Morris JK, Garne E, Wellesley D, et al. Major congenital anomalies in babies born with Down syndrome.

14th International Conference on Pediatrics & Neonatal Healthcare
March 11-12, 2020 | Zurich, Switzerland

Author Citation: Takako Takano, *Sex differences in congenital heart disease in Japanese Down Syndrome*, Pediatrics Healthcare 2020, 14th International Conference on Pediatrics & Neonatal Healthcare, Zurich, Switzerland, March 11-12, 2020