Impact of national prenatal screening program on prenatal detection of Transposition of the Great Arteries over a 10-Year Period: Improved, but not enough

Introduction

Transposition of the great arteries (TGA) is one of the most common cyanotic congenital heart defects requiring early neonatal surgery. Earlier research has demonstrated an increase in prenatal detection of simple TGA, improved survival, and quality of life in the current era.

We aimed to evaluate the impact of the introduction of a systematic national prenatal screening program on prenatal detection of simple TGA (+/- ventricular septal defect) and to evaluate the regional differences in total prevalence in the five university districts in an 11 – year national cohort from Finland.

Methods

The systematic national prenatal screening was introduced in January 2007 and has been fully implemented from January 2010 onwards. We compared prenatal detection rates of TGA in three time periods: Before the introduction of systematic ultrasound screening 2004–2006, transition period 2007–2009 and screening period 2010–2014. Data on births (all births n=651 970) and pregnancy terminations with the diagnosis of simple TGA during 2004–2014 were assessed from five national registers (Register of Pediatric Cardiac Surgery, Register of Congenital Malformations, Register of Induced Abortions, Medical Birth Register and Cause of Death Register).

Results

There were 144 cases with simple TGA (131 live births, one stillbirth and 12 terminations of pregnancy). National total prevalence of simple TGA was 2.21/10 000 births (western part of Finland 1.78/10 000, southern 2.26/10 000, northern 2.28/10 000 and eastern 3.13/10 000). There was a significant difference in the total prevalence between eastern and western part of the country (p=0.020).

The overall prenatal detection rate was 28.5% (41/144). The proportion of prenatally diagnosed cases increased significantly from 14.0% (2004–2006), 22.0% (2007–2010), to 43.3% (2010–2014) (p=0.001). However, the impact of the screening program had significant regional variation (p<0.05).

The random diagnoses (diagnosed outside the screening program) decreased from 50% to 0% (p=0.001) and delayed diagnoses (diagnosis after 24 weeks control ultrasound due to abnormality or suboptimal views in screening) from 50% to 15% (p=0.034).

Conclusions

Introduction of the national prenatal screening program has significantly increased the prenatal detection of TGA (14.0%–43.3%), but the impact of the screening program varied inside the country. Interestingly, even genetic isolate country such as Finland has significant regional variation in prevalence of TGA warranting for future studies.