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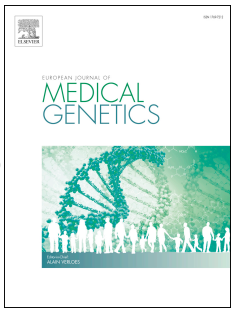

The impact of prenatal diagnosis on congenital anomaly outcomes: Data from 1997 to 2016

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Title: The impact of prenatal diagnosis on congenital anomaly outcomes: data from 1997 to 2016

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Abstract

The term prenatal diagnosis comprises a variety of techniques aimed to determine the health and condition of the embryo or foetus. Its main goal is to identify at an early stage of the pregnancy, if possible, malformations or other conditions that could increase the risk of a negative outcome in the pregnancy. The aim of this study was to assess the impact of prenatal diagnosis in Portugal in pregnancies with congenital anomalies. A cross sectional study was implemented using data for the years 1997 to 2016 from the Portuguese registry of congenital anomalies (RENAC), a population-based registry that follows EUROCAT guidelines. Analysis was restricted to malformations that are potentially detectable by prenatal diagnosis. The effect of prenatal diagnosis on outcome (death vs live birth) was estimated using a regression model. Main results indicate that prenatal diagnosis was performed in 56.1% (n=7605) of all registered cases. At least one malformation was detected for the first time through ultrasound (47.4%), invasive tests (5.6%) and other tests (2.2%). When analysed severe CA, 54.2% was detectible by prenatal ultrasound distributed as follows: 17.4% were diagnosed before 14 weeks of gestation, 47.6% between 14 and 23 weeks and 35.0% with 24 or more weeks of gestation. TOPFA was the option for 21.3% of these CA.

Over the 20 years of analysis, there was a statistically significant increase trend in the detection rate of congenital anomalies through prenatal diagnosis compared to detection at birth or after birth ($p<0.001$).

After adjusting for confounding (year, maternal age, presence of more than one malformation), prenatal diagnosis was associated with more severe outcomes (TOPFA, 40.3%; Death 3.5%) and increased the risk of the pregnancy ending in foetal death (OR=2.56; 95%CI=2.06-3.18). These results are in accordance that more severe anomalies are more easily detected prenatally.

Considering the results, it is important to raise awareness about the importance of pregnancy planning and preventing the risk factors more associated with CA. More information about prognosis for children with congenital malformations is important for parents and health professionals after prenatal detection.

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