The evolution of prenatal diagnosis in the early detection of congenital anomalies: data from 1997 to 2016

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BACKGROUND

- Prenatal diagnosis comprises a variety of techniques aimed to determine the health and condition of the embryo or foetus.
- It has been used in clinical practice for more than 40 years, but it was during the last two decades that advances in non-invasive tests such as ultrasound technology, and invasive techniques such as amniocentesis or chorionic villus sampling for pregnancies at increased risk for chromosomal anomalies or genetic diseases have started to be used more frequently.
- The Portuguese prenatal surveillance programme advise ultrasound screenings, in the first trimester of pregnancy in combination with the screening test, and between 20 - 22 weeks of pregnancy. Other tests are offered in pregnancies with an increased risk (1).

OBJECTIVE

The aim of this study is to assess the evolution of prenatal diagnosis in the detection of congenital anomalies (CA).

METHODS

A cross sectional study was implemented using data collected between 1997 and 2016 by the Portuguese registry of CA (RENAC).

RENAC (2) is a population based registry, full member of EUROCAT since 1996 and follows the European registry guidelines (3).

For this study a case was defined with at least one CA potentially detectible by prenatal diagnosis (Table 1).

Descriptive analysis was performed using absolute and relative frequencies and bivariate analysis was conducted using chi-square statistics.

RESULTS

The analysis included 13,566 cases reported with at least one of the studied CA.

There was an statistically significant increase in the detection of CA through prenatal diagnosis compared to detection at birth or after birth (Figure 1, Table 2, p<0.001).

In addition, there was an increase of cases detected during pregnancy from 52.1% (1997-1999) to 62.9% (2009-2016) especially in cases detected before 14 weeks (7.9% to 28.9%).

Comparing the same periods of time the results also show a range of ultrasound screening from 27% to 55.8% and a decrease in invasive tests from 18% to 3%. This tendency was statistically significant (p<0.001).

CONCLUSIONS

The data show a positive effect on the percentage of cases with CA detected during pregnancy. These results show the importance of extending prenatal tests to the all pregnant women and not only to those with specific risk gestations.

REFERENCES: