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.

Table 1. List of congenital anomalies included in the study

Nervous system: Anencephalus and similar; Hydrocephalus; Spina Bifida; Holoprosencephaly.
Severe congenital heart defects: Transposition of great vessels; Coarctation of aorta; Tetralogy of Fallot; Hypoplastic left heart; Single ventricle; Common arterial truncus; Tricuspid atresia and stenosis; Ebstein's anomaly; Aortic valve atresia/stenosis; Pulmonary valve atresia/stenosis.
Non severe congenital heart defects: Ventricular septal defect; Atrial septal defect.
Digestive system: Diaphragmatic hernia
Abdominal wall defects: Gastroschisis; Omphalocele.
Urinary: Bilateral renal agenesis; Renal dysplasia; Congenital hydronephrosis.
Limb: Limb reduction defects; Club foot - talipes equinovarus.
Oro-facial clefts: Cleft lip; Cleft palate with or without cleft lip.
Chromosomal: Trisomy 21; Trisomy 18; Trisomy 13.

RESULTS

The analysis included 13566 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).

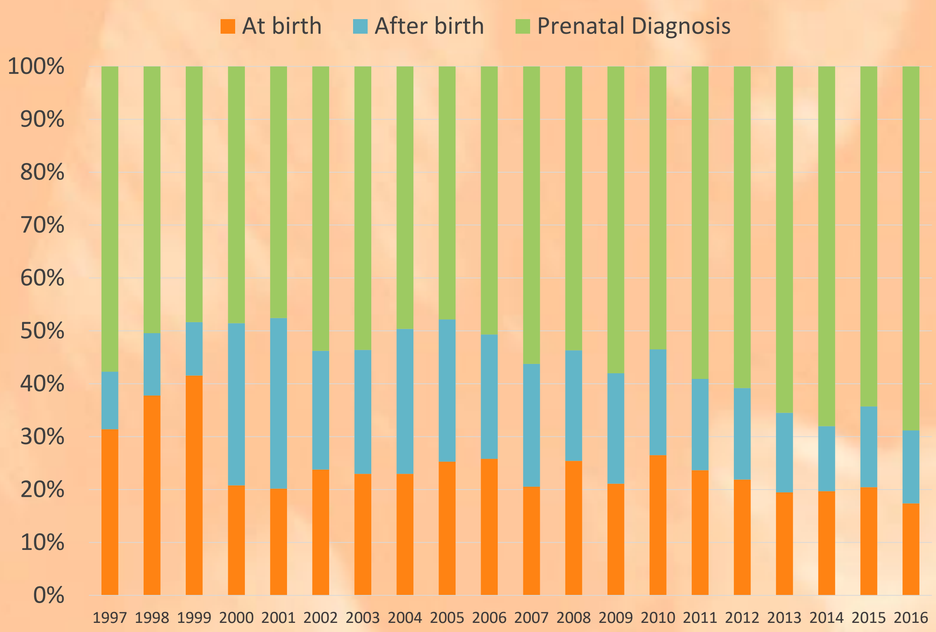

Figure 1. Time of diagnosis of congenital anomalies from 1997 to 2016

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 18.9% to 3%. This tendency was statistically significant (p<0.001).

Table 2. Evolution of prenatal diagnosis, gestational age at discovery and diagnosis test performed from 1997 to 2016

		1997-1999		2000-2008		2009-2016		
		n	%	n	%	n	%	*p-value***
Prenatal diagnosis	No	852	47.9	2980	49.1	2116	37.1	<0.001
	Yes	926	52.1	3096	50.9	3583	62.9	
GA* at discovery	<14 week	39	7.9	249	9.9	847	28.9	<0.001
	14-23 week	285	58.2	1192	47.3	1380	47.2	
	>=24 week	166	33.9	1081	42.9	697	23.8	
Prenatal tests	Ultrasound	486	27.2	2,78	45.8	3,168	55.6	<0.001
	Invasive tests	338	18.9	246	4.05	173	3.03	
	Other tests	15	0.84	33	0.54	256	4.50	
	No prenatal tests	950	53.1	3017	49.7	2104	36.9	

*GA - gestational age; **p-value of chi-square test for comparison of independent variables

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.

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