Measuring improvement of prenatal diagnosis efficiency in Tuscany.

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Introduction and Objective

Congenital anomalies represent a major cause of infant mortality, childhood morbidity and long-term disability, with a prevalence at birth of 3-5%. The Tuscany Registry of Congenital Defects (RTDC) was set up in 1992 to guarantee full registration of cases in the Region.

Efficiency of ultrasound screening in detecting foetal anomalies has considerably improved over the past 20 years and gestational age at the time of diagnosis of birth defects is considered a crucial indicator to provide better quality health care services.

The Tuscany Registry of Congenital Defects has assessed whether gestational age at diagnosis of selected congenital defects has decreased over years in Tuscany (Italy).

Methods


Defects were selected among those more likely to be prenatally diagnosed: anencephaly, spina bifida, encephalocoele, holoprosencephaly, omphalocele, gastroschisis, hypoplastic left heart, tetralogy of Fallot, transposition of great vessels, common atrioventricular canal, single ventricle and bilateral renal agenesis.

Diagnostic efficiency was evaluated comparing mean gestational age differences (MGADs) at prenatal diagnosis among each period of time by univariate ANOVA with Bonferroni test, the regression being performed to evaluate linear trend.

Results

An overall decrease of mean gestational age at diagnosis emerged for almost all selected defects.

Statistically significant mean gestational age differences for gestational age at diagnosis were found for spina bifida and hypoplastic left heart between the 2003-2008 and 1992-1996 periods (MGAD = -2.86 weeks p=0.04; MGAD = -5.36 weeks p=0.07 respectively).

A mean decrease of 1.43 weeks, 1.74 weeks, 1.82 weeks emerged for almost all selected defects.

Results showed differences between mean gestational age at diagnosis before and after 1997 for holoprosencephaly (p=0.07) with a mean decrease of 6.25 weeks between the two periods.

Conclusions

Results of the study show improved efficiency of prenatal diagnosis in Tuscany Region over 1992-2008. Although improvement over time is not always statistically significant, earlier gestational age at diagnosis for almost all selected defects emerged.

Findings of the study indicate that measurement of the detection time of congenital defects may be a useful performance indicator and may lead to secondary prevention services for prenatal diagnosis.