Environmental factors in the etiology of esophageal atresia and congenital diaphragmatic hernia: results of a case-control study.

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Abstract

BACKGROUND: Esophageal atresia with or without tracheoesophageal fistula (EA/TEF) and congenital diaphragmatic hernia (CDH) are severe congenital anomalies. Their etiologies are mostly unknown and are thought to be multifactorial. No specific environmental factors have consistently been described as risk factors.

METHODS: In a study conducted during the years 2000 to 2004 in a pediatric surgical referral center in the Netherlands, parents of children with EA/TEF or with CDH of the Bochdalek type and parents of a group of children without major birth defects filled out a questionnaire about possible exposure to environmental risk factors during the period from 1 month before conception to the end of the first trimester of pregnancy. Children with chromosomal anomalies were excluded. Questionnaires were returned for 47 out of 64 cases (73%) with EA/TEF, for 63 out of 77 cases (82%) with CDH, and for 202 out of 243 controls (83%). In EA/TEF, maternal age was borderline significantly higher than in controls (32.2 vs. 30.6 years, p = .05). Contact with herbicides or insecticides was associated with EA/TEF in univariate analysis (OR 2.0; 95% CI: 1.0-4.1) and in multivariate analysis, although of borderline significance. In univariate analysis, CDH was significantly associated with maternal use of alcohol (OR 2.9; 95% CI: 1.6-5.2).

RESULTS: Questionnaires were returned for 47 out of 64 cases (73%) with EA/TEF, for 63 out of 77 cases (82%) with CDH, and for 202 out of 243 controls (83%). In EA/TEF, maternal age was borderline significantly higher than in controls (32.2 vs. 30.6 years, p = .05). Contact with herbicides or insecticides was associated with EA/TEF in univariate analysis (OR 2.0; 95% CI: 1.0-4.1) and in multivariate analysis, although of borderline significance. In univariate analysis, CDH was significantly associated with maternal use of alcohol (OR 2.9; 95% CI: 1.6-5.2).

CONCLUSIONS: We found a significant association between maternal alcohol use around the time of conception and CDH. A possible explanation might be the effect of alcohol on the retinoic acid pathway. An association was found between contact with herbicides or insecticides and EA/TEF.

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Comment in

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