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Tracheo-esophageal anomalies in siblings

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Summary

Two cases of consecutive siblings born with tracheoesophageal fistula are presented. The literature pertaining to this subject is reviewed and briefly discussed.

Key words: *Tracheoesophageal fistula, siblings*

Introduction

The cause of esophagel atresia with or without tracheoesophageal fistula (TEF) is unknown [12]. Familial occurrence of congenital esophageal atresia and TEF have been reported infrequently [10,14]. Up to now a clear genetic influence has not been demonstrated [6,17]. In this report detailed description of two cases of esophageal atresia with TEF in two consecutive siblings have been presented.

Case Report

A baby boy, second child of the family, was born full-term by secondary ceserean section on July 1989. Birth weight was 3450 g. Initially he appeared healthy in nursery, but after a short time, he could not swallow and was observed to produce large amounts of frothy sputum. An attempt to place a nasogastric tube was unsuccessful. Chest x-ray showed right upper lobe atelectasis and a radiopaque catheter curled up in the upper esophageal pouche. A diagnosis of esophageal atresia and TEF was made.

During the examination, the infant was icteric and well developed. The pulse rate was 140 per minute, blood pressure was 80 mmHg systolic and rectal temperature 37°C. There were no abnormalities of the head, face, palate, trunk or extremities. The abdomen was mildly distended and soft. There were no palpable masses or enlarged viscera. Genitalia were normal, and a drop of meconium was visible at the anus. Total bilirubin was 11.3 mg/dl, indirect bilirubin was 10.8 mg.

The first boy of this family has been born by ceserean section in the same hospital with esophageal atresia and TEF in 1985. He had been operated in another hospital and a primary anastomosis was performed. Chromosome studies carried out in all members of the family. No pathological finding was oloserved.

Discussion

Esophageal atresia occurs approximately once in every 4000 live births [6,15]. The error in development is thought to occur during the third or fourth week of fetal life [9]. While the empirical change of a single malformation recurring in immediate relavites in said to be 3 % to 5 %, this does not seem to be true in TEF [1].

The cause of esophageal atresia with or without fistula is unknown. Several interesting facts are, however, established. The occurrence of other anomalies (particularly the VACTERL association) with TEF suggest a teratogen or genetic makeup that may influence several systems; the cardiovascular and gastrointestinal systems are most ofter involved [1,7,10,16].

Cases have been reported in the same families on four occassions; one family had three affected siblings, brothers, sisters, cousins and brother-sister combinations with TEF [4,5,6].

Esophageal atresia was reported in both identical

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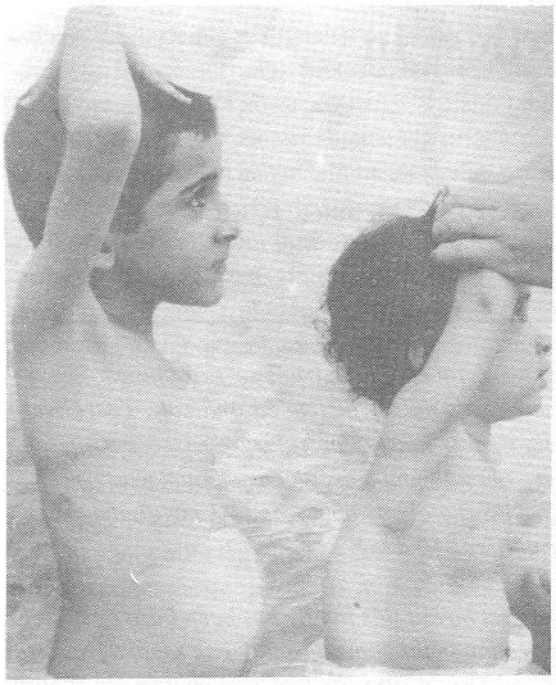

Figure 1. Lateral view of two brothers showing the site of operation.

twins [1,8]. But there are many more instances of one twin having the anomaly and one spared [3,9]. German and associates [8] reported an incidence of 9 % of twining in 102 patients with esophageal atresia (The incidence of twining ise 2.5 % among all live births). According to Haight [11] esophageal anomalies arise from a developmental disruption rather than being genetically transmitted disease. Chen and his associates [2] concluded that embryologic and environmental factors play a major role in the development of esophageal atresia and the cause of the defect might be bultifactorial, but heritability of the defect is low. Ozimek and sassociates [15] reported a cyclical incidence of the anomaly, suggesting the possibility of a causative infectious agent, such as hepatis. In an epidemiliologic study by Ingalls and Prindle [13], the mothers of children with the deformity were occasionally older, the children were illegitimate with twice the expected frequency, and placental defects were four times as common as expected. No associated abnormalities such as infection, placental defect or chromosomal pathology were found in our cases.

When a family history of TEF exists, the pediatrician should be quick to rule out an esophageal atresia by inserting a tube under fluoroscopic control into the proximal esophagus [14].

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