Familial Tracheoesophageal Atresia

*Alexandra Anga* and B. A. Animashaun

1Division of Neonatology, Department of Paediatrics, Lagos State University Teaching Hospital (LASUTH), Nigeria

2Division of Cardiology, Department of Paediatrics, Lagos State University College of Medicine (LASUCOM), Nigeria

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*Corresponding author:* Alexandra Anga, Division of Neonatology, Department of Paediatrics, Lagos State University Teaching Hospital (LASUTH), Ikeja, Lagos, Nigeria.

Introduction

Oesophageal Artesia most times accompanied by tracheoesophageal fistula is not an uncommon clinical condition. It occurs in 1 in 2,500 to 3,000 live births and there is little evidence to support significant geographical or secular variation in the incidence. The majority of cases of Oesophageal Artesia are sporadic/non syndromic. Familial/syndromic cases of oesophageal Atresia are extremely rare representing less than 1% of the total [1]. Survival is directly related to birth weight and to the presence of a major cardiac defect [1]. Infants weighing over 1,500 gram and having no major cardiac problem should have a survival rate of near 100% [1,2].

Case Report

We report a case of a male child born at 41 weeks gestation after an uneventful pregnancy to a 36 year old woman, through an uncomplicated Caesarean section. The birth weight was 3,215g. There was however a family history of Oesophageal Artesia with a distal fistula in the baby’s first cousin. The Apgar scores were 9 at one minute and ten at five minutes. Meconium was passed within six hours of delivery. A slight increase in whitish oral secretions was noted. After two hours of life, the baby developed respiratory distress despite the fact that he had not yet been fed. The respiratory rate was 96 breaths per minute; there was reduced intensity of breath sounds at the lung bases and generalised rhonchi.

![Figure 1: Initial diagnosis.](image)

An initial diagnosis was made of aspiration pneumonitis (Figure 1). After an initial turbulent period the baby’s clinical condition improved. On the fourth day of life repeated attempts to pass a nasogastric tube for feeding failed. Intraoesophageal suction continued to yield copious aspirates. A tube containing light barium was passed and a plain radiograph of the neck and chest taken (Figure 2). This demonstrated a blind proximal oesophageal pouch. A plain abdominal radiograph showed abundant gastric and intestinal gas (Figure 3). Bronchoscopy subsequently confirmed oesophageal Atresia. Clinical evaluation and an echocardiogram excluded congenital heart disease. The baby underwent primary surgical repair on the eighth day of life and has since recovered.
TOF remains a prime consideration in a neonate who develops these as soon as possible [1,2,4]. Oesophageal Atresia with an associated malformation and effort should be made to detect should be noted that fifty per cent of affected babies will have and the absence of cardiac and other associated anomalies. It to a good outcome in babies of weights above 1500 grammas and TOF has significantly improved over the last decade [4].

The presence of an associated defect negatively impacts on the outcome. Our patient did not have any congenital cardiac abnormality. One major feature of interest in the index case is the fact that a paternal first cousin also had tracheo-oesophageal fistula. This underlines the fact that although most cases are sporadic, familial cases do exist [3], we wonder if this could be a case of familial Tracheoesophageal fistula. The overall data about Oesophageal atresia in twins reveal discordance in the great majority with a concordance rate as low as 2.5% [4]. The recurrence risk of the anomaly in siblings of an affected child calculated from data from familial studies is comparable with a recurrence risk of a multifactorial condition (1%). And the distribution of the anomaly in first and second degree relatives does not fit into a monogenic mode [3].

Conclusion

These data suggest that in most cases aetiopathological factors are non genetic. No pedigree so far has been demonstrated with direct transmission over three generations [2]. There are however, well-defined instances where genetic factors clearly play a role [1]. Recently three genes have been identified in humans with a role in OA. Feingold syndrome (N-MYC), Anopthalmia – OA [1]. Recently three genes have been identified in humans with a well-defined role in Tracheoesophageal malformations in which there is a blind proximal oesophageal pouch and a distal fistulous connection with the trachea. Tracheoesophageal malformations are often associated with other congenital abnormalities mostly cardiac like VSD, PDA or Fallots Tetralogy and may be a component of VATER association.

Prompt recognition, appropriate clinical management to prevent aspiration and swift referral to an appropriate tertiary specialist centre will result in a significant improvement in rates of morbidity and mortality. Antenatal diagnosis however allows the delivery centre to be optimised thus improving the outcome. Improvement in the survival of high risk cases is related to better post operative care [6] Long term follow up is essential because of the ongoing morbidity of the patients [5-12].

References
