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ORIGINAL ARTICLE

Oesophageal atresia with cleft lip and palate: a marker for associated lethal anomalies?

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Abstract An adverse association between oesophageal atresia (OA) and cleft lip-palate (3% incidence) has been reported. The present study analyses outcomes of this rare association at a UK paediatric surgical centre. Hospital charts of newborns diagnosed with OA were reviewed. Demographics, associated anomalies and prognostic classification (after Spitz 1994) were recorded. Mortality rates and causes of death were examined in OA babies with cleft lip-palate. Of 152 patients treated for OA, five babies (3%) had cleft lip-palate. All of these newborns had common variant OA-TEF and were Spitz group II category. Deaths occurred in 3 of 5 patients (60%) in the OA-cleft group compared to only 8 of 147 patients (5%) without clefts ($p < 0.005$; Fisher's exact test). OA-cleft non-survivors succumbed to tetralogy of Fallot ($n = 2$) and trisomy 18 ($n = 1$; treatment withdrawn). Both survivors with cleft lip-palate had features of the VACTERL sequence: one baby also had Goldenhaar syndrome, the other aortic coarctation. These children now attend mainstream school. Although high-quality survival is possible in OA with cleft lip-palate, this rare phenotype is associated with a substantially decreased survival. Rather than causing death directly, the combination of OA and cleft lip-palate appears to be a marker for further lethal anomalies.

Keywords Oesophageal atresia · Tracheo-oesophageal fistula · Cleft lip · Cleft palate

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Introduction

Cleft lip and palate (CL-P) are reported in around 2–3% of newborns with oesophageal atresia [1–4]. An adverse association between oesophageal atresia (OA) and CL-P has been documented in a single study [4]. In light of these findings we sought to analyse the survival and outcome of babies with OA and CL-P at a specialist UK paediatric surgery centre.

Methods

Hospital case records of all newborns admitted with a diagnosis of OA were examined. Demographics, associated anomalies and the Spitz classification were recorded. Mortality and causes of death were further studied in OA babies with associated CL-P.

Results

A total of 152 patients were admitted to the regional neonatal surgical unit with a diagnosis of OA during the 15 year period 1980–2004. Five (3%) babies (2 male, 3 female) were identified with associated CL-P anomalies. The mean birth weight of this cohort was 2.23 kg (range 1.57–2.8). All infants with CL-P birth defects had common variant OA with a distal tracheo-oesophageal fistula (TEF). There were no cases of 'long gap', H type fistula or pure OA in this patient group. Five infants were identified as Spitz group 2 (<1.5 kg or major cardiac anomaly) according to the classification proposed by Great Ormond Street Children's Hospital [1]. All of these babies had a co-existent cardiac anomaly (tetralogy of Fallot $n = 3$,

Table 1 Details of patients with oesophageal atresia and associated cleft lip-palate anomalies

Patient no.	Gender	Birth weight	Orofacial anomaly	Type of OA	Associated anomalies	Syndrome	Spitz class	Cleft repair	Outcome (cause of death)
1	M	2.80	Bilateral cleft lip, alveolus and palate	OA distal TOF	Aortic coarctation, 2 absent ribs, T10 hemi-vertebra, right leg weakness	VACTERL	2	Yes	Alive 4 years
2	F	2.30	Left sided cleft lip and palate	OA distal TOF	Patent ductus arteriosus, ventricular septal defect, dextrocardia, facial dysmorphism	VACTERL	2	No	Died at 5 days (cardiac failiure)
3	F	2.10	Right sided cleft lip, alveolus and palate	OA distal TOF	Dextrocardia, hemi-facial microsomia	VACTERL, Goldenhaar syndrome	2	Yes	Alive 10 years
4	M	2.36	Bilateral cleft lip and palate	OA distal TOF	Tetralogy of Fallot, abnormal pul arteries, large PDA, extra digit left hand, micropenis	CHARGE	2	Yes	Died at 13 months (cardiac failure)
5	F	1.57	Left sided cleft lip and alveolus	OA distal TOF	Tetralogy of Fallot	Trisomy 18	2	No	Died at 16 days (trisomy 18)

dextrocardia, VSD + PDA $n = 1$, coarctation of aorta $n = 1$). Data for these infants are summarised in Table 1. Four newborns had a primary oesophageal anastomosis with corrective division and repair of the TEF. Three (60%) babies with facial clefts died following repair of their OA-TEF, compared to 8/147 in the non-cleft group ($p < 0.005$; Fisher's exact test). Of these three deaths, two newborns had major cardiac anomalies (patient no. 2 had a PDA and VSD with dextrocardia, and patient no. 4 had tetralogy of Fallot with a large PDA and an abnormal pulmonary arterial system) leading to fatality, and one child had a lethal chromosomal anomaly—Edward's syndrome (trisomy 18) with tetralogy of Fallot—resulting in withdrawal of active treatment. Both survivors with CL-P (patient numbers 1 and 3 on Table 1) had features of the VACTERL sequence: one of these babies also had Goldenhaar syndrome, and the other aortic coarctation. Both children enjoy a good quality of life and are performing well at mainstream school.

Discussion

In this study we report a 3% incidence of CL-P associated with OA-TEF in a regional UK neonatal surgical centre treating >10 new index cases annually. This observation concurs with findings recently reported from Amsterdam and Great Ormond Street, London [1–4]. Is CL-P a marker of poor outcome in babies with OA-TEF as suggested by Spitz and colleagues? [4]. Our study would seem to indicate that cardiac and chromosomal anomalies are a better indicator for lethality. This is in agreement with a critical analysis highlighted by Deurloo and Aaronson from the Netherlands [3]. Fatalities in this 'high risk' group are closely linked to the severity of the underlying congenital heart disease and lethal chromosomal anomalies. The incidence of cardiac problems in CL-P patients is variably reported to range between 1.3 and 15% [5–7]. Survivors in our study enjoy a good quality of life and attend mainstream schools. This modest success has been attributed to multidisciplinary management linking several specialist teams—intensivists, neonatologists, surgeons, respiratory paediatricians and skilled nursing staff. In the assessment of a newborn with OA-TEF and facial cleft, clinicians must maintain a high index of suspicion of associated congenital heart disease. This study reinforces the requirement for complete pre-operative cardiac assessment in these complex newborns to guide treatment and decision making. Counselling following diagnosis in the antenatal and postnatal period should carefully reflect these issues.

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