Tracheoesophageal Fistula with Esophageal Atresia with Absent External Ear: A Unusual Association

Vijai D. Upadhyaya · Ajay N. Gangopadhyay ·
Punit K. Srivastava · Zaheer Hasan · Shiv P. Sharma

Received: 5 January 2009 / Accepted: 23 March 2009 / Published online: 1 May 2012
© Association of Surgeons of India 2012

Abstract Esophageal atresia with tracheoesophageal fistula (TEF with EA) is a common disease which is associated with many other congenital abnormalities. The most common association is with VACTERL syndrome. Various types of ear malformations have been reported in CHARGE syndrome associated with EA and TEF. However, absence of external ear has not been described which is being reported in this article. We could not find this association even on extensive search of literature.

Keywords Esophageal atresia · Tracheoesophageal fistula · Ear deformity · Absent pinna

Introduction

Esophageal atresia and tracheoesophageal fistula (EA and TEF) remain the epitome of neonatal surgery with its estimated live birth incidence of 1 in 3,570 to 1 in 4,500. Esophageal atresia (EA) is often associated with anomalies of other systems. The anomaly may be obvious or hidden; it may be simple or complicated and it may or may not be life-threatening. The embryologic missteps leading to esophageal atresia are associated with varying components of the malformation constellation termed VACTERL, CHARGE association and ‘SCHISIS’ syndrome. The cardinal features are coloboma, heart anomalies, choanal atresia, retardation of growth, genital and ear anomalies clinically define CHARGE syndrome.

No single feature is universally present or sufficient for diagnosis, and the severity is variable. Other frequently occurring features of significance include: characteristic face and hand dysmorphism, hypotonia, urinary tract anomalies, orofacial clefting, deafness, and dysphagia and tracheoesophageal anomalies. The clinical diagnosis requires the presence of at least 3 cardinal features, or 2 cardinal features plus at least 3 associated minor characteristics. Ear deformity in CHARGE association, involves internal ear with deafness but in present case internal ear was normal and there was almost absence of pinna. Since our patient was not have any neurological disorder or facial palsy or deafness hence it cannot be included in CHARGE association. We are presenting the cases of esophageal atresia with tracheoesophageal fistula with unilateral absence of pinna with normal internal ear without other obvious congenital anomaly or neurological abnormality.

Case Report

A 2.5 kg male baby was admitted to hospital on the day one of life with drooling of saliva and absent unilateral pinna. The baby was restless without cyanosis and after oropharyngeal suction the catheter was not passed beyond 8 cm from the upper alveolus. Thorough examination of the baby revealed no other associated congenital anomalies except absence of the right side external ear (Fig. 1).

The nasogastric tube was found to be coiled up in the upper chest at the level of second thoracic vertebra on X-ray chest. The baby was adequately resuscitated and the tracheoesophageal fistula was ligated and an end to end esophago-esophageal anastomosis was performed by an extrapleural

V. D. Upadhyaya (✉) · A. N. Gangopadhyay · P. K. Srivastava ·
Z. Hasan · S. P. Sharma
Pediatric Surgery,
IMS,
BHU, Varanasi, India
e-mail: upadhyayavj@hotmail.com

Fig. 1 Clinical photograph showing almost absent pinna

right thoracotomy. Postoperative course was uneventful. A patient was investigated thoroughly but no other anomalies

Fig. 2 CT scan showing normal middle ear

associated with CHARGE association. Patients did well in post-operative period and waiting for ear surgery. On follow up at age of 2 year patient was subjected to CT scan which was suggestive of normal internal ear (Fig. 2). Now patient is waiting for cosmetic surgery.

Discussion

EA and TEF result from an early disturbance in the organogenesis, the incidence of which varies between 40 and 55% [1–3]. These anomalies are most common in pure EA (55–60%) and least common in H-type TEF (25–30%) [3]. Cardiovascular anomalies occur most frequently (11–49%) followed by genitourinary (24%) [4], gastrointestinal (24%) [5] and skeletal (13%) [6] anomalies. Associated neurological anomalies include neural tube defects (2.3%), hydrocephalus (5.2%), holoprocencephaly (2.3%) and anophthalmia or microphthalmia (3.7%) [7]. Other anomalies are choanal atresia (5.2%), facial cleft (7.2%), abdominal wall defects (4.3%) and diaphragmatic hernia (2.9%) [7]. In 1973 Quan and Smith [8] suggested a broad spectrum of associated malformations that might or might not appear together and are associated with EA. They arranged this association into the acronym, VATER: vertebral defects, anal atresia, tracheoesophageal fistula, esophageal atresia and renal defects. As the phenotype expanded, the acronym was changed to VACTERL association which included vertebral, anorectal, cardiac, tracheoesophageal, renal and radial limb abnormalities [9]. Although infrequently EA has also been reported in SCHISIS association (omphalocele,

Fig. 3 CT scan showing normal inner ear

neural tube defects, cleft lip and palate and genital hypoplasia) [7, 10]. Pagon et al. ascertained and reported additional children with either choanal atresia or coloboma and associated characteristic malformations, and coined the acronym CHARGE association (Coloboma, Heart Defect, Atresia Choanae, Retarded Growth and Development, Genital Hypoplasia, Ear Anomalies/Deafness) [11]. Facial nerve and renal anomalies are considered common adjuncts to this spectrum [12]. The ear deformities associated with this association are: external ear lops or cup shaped ear, middle ear deformity and mixed deafness. In present case there is total absence of external ear. Despite the extensive literature search we could not find any association of this type of anomaly with esophageal atresia. This case was presented because of its rarity and its dissimilarity from the CHARGE syndrome (Fig. 3).

References

1. Ein SH, Shandling B, Wessen D, Filler RM (1989) Esophageal atresia with distal tracheoesophageal fistula: associated anomalies and prognosis in 1980s. *J Pediatr Surg* 24:1055–1059
2. Holder TM, Ashcraft KW, Sharp RJ, Amoury RA (1987) Care of infants with esophageal atresia, tracheoesophageal fistula and associated anomalies. *J Thorac Cardiovasc Surg* 94:828–835
3. Kluth D (1976) Atlas of esophageal atresia. *J Pediatr Surg* 11:909–919
4. Beasley SW et al (1992) Urinary tract abnormalities in association with EA frequency, significance and influence on management. *Pediatr Surg Int* 7:94
5. Spitz L, Kiely EM, Brereton RJ, Drake D (1993) Management of esophageal atresia. *World J Surg* 17:296–300
6. German JCC, Mahour GH, Woolley MM (1976) Esophageal atresia and associated anomalies. *J Pediatr Surg* 11:299
7. Harris J, Kallen B, Robert E (1995) Descriptive epidemiology of alimentary tract atresia. *Teratology* 52:15–29
8. Quan L, Smith DW (1973) The VATER association. *J Pediatr* 82:104
9. Baumann W, Greinacher I, Emmrich P, Spranger J (1976) Vateroder vacterl syndrome. *Klin Padiatr* 188:328–327
10. Chittmittrapap S, Spitz L, Kiely EM, Brereton RJ (1989) Oesophageal atresia and associated anomalies. *Arch Dis Child* 64:364–368
11. Pagon RA, Graham JM Jr, Zonana J, Yong S (1981) Coloboma, congenital heart disease, and choanal atresia with multiple anomalies: CHARGE association. *J Pediatrics* 99:223–227
12. Morgan D, Bailey M, Phelps P, Bellman S, Grace A, Wyse R (1993) Earnose-throat abnormalities in the CHARGE association. *Arch Otolaryngol Head Neck Surg* 119:49–54