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Antenatal Diagnosis of Esophageal Atresia

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Abstract **Case Report**

Oesophageal atresia (OA) encompasses a group of congenital anomalies comprising of an interruption of the continuity of the oesophagus with or without a persistent communication with the trachea. OA occurs in 1 in 2500 live births. We report the case of a 33 year old patient referred to our unit for severe Polyhydramnios at 24 week of amenorrhea. Ultrasound examination revealed the absence of stomach bubble. During fetal swallowing we observed a pouch at the upper level of the neck not vascularized at doppler. There was no other associated fetal abnormality and no chromosomal abnormality. We discussed the preened post-natal diagnosis of esophageal atresia, associated malformations should be seeded and management of esophageal atresia.

Keywords: Oesophagial Atresia, Pough Sign, stomach bubble, polyhydramnios.

INTRODUCTION

Oesophageal atresia encompasses a group of congenital anomalies comprising an interruption of the continuity of the oesophagus combined with or without a persistent communication with the trachea.

The first clearly documented case and confirmed at post mortem examination was recorded by Thomas Gibson in 1697 [1], the etiology is multifactorial and remains unknown.

We have reported the case of an esophageal atresia diagnosed prenatally in our obstetrics and gynecology department I at Hassan II CHU in Fez.

OBSERVATION

We report the case of a 33-year-old patient, without significant pathological antecedent, gravidia 4 para 3, with three normal vaginal deliveries without incident. The patient was referred to our unit at 24 weeks of amenorrhea with severe Polyhydramnios. Obstetrical ultrasound examination revealed the absence of stomach bubble (Figure-1) with an abdominal circumference below the 10th percentile. During fetal swallowing we observed a pouch at the upper level of the neck not vascularized at doppler (Figure 2 & 3). There was no other associated fetal abnormality and no chromosomal abnormality.

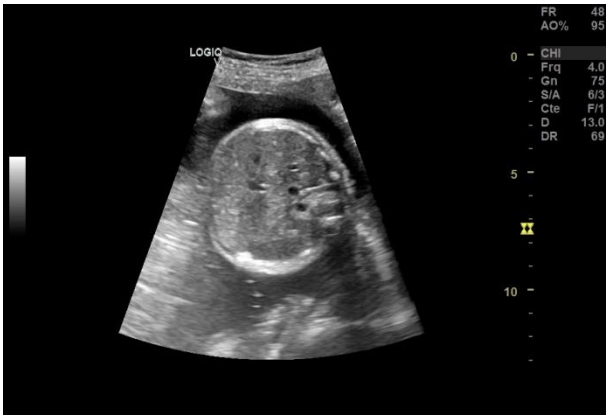

Fig-1: Transverse view of fetal abdomen showing the absence of stomach bubble

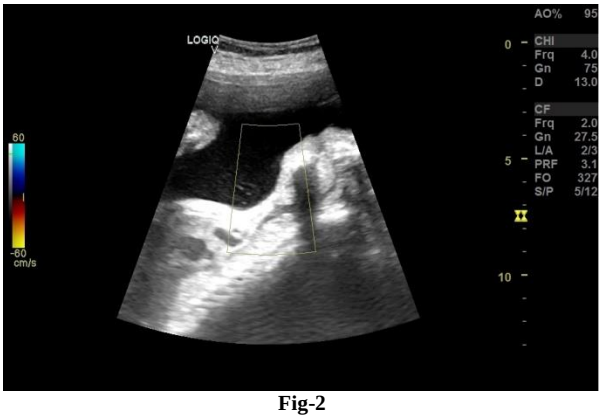

Fig-2

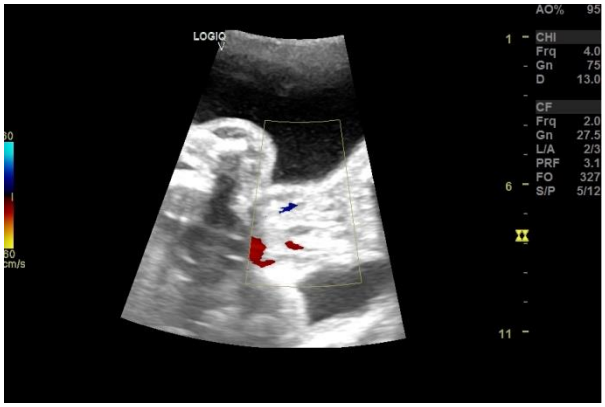

Fig-3: Ultrasound and color doppler images showing the pouch sign in fetal neck

DISCUSSION

Oesophageal atresia is a relatively common congenital malformation occurring in one in 2500–3000 live births. The majority of cases of oesophageal atresia are sporadic/non-syndromic. Familial/syndromic cases of oesophageal atresia are extremely rare, representing less than 1% of the total. Oesophageal atresia is 2 to 3 times more common in twins than the total population [2].

The first clearly documented case and confirmed at post mortem examination was recorded by Thomas Gibson in 1697 [1], then Thomas Hill in 1840 reported the second case of esophageal atresia but the first one associated with an anal anomaly [3].

Five major types of tracheoesophageal abnormalities was first described by Vogt in 1929 [4], still used today, and then have been modified by Ladd in 1944 [5], Gross in 1953 [6] and Kluth in 1976 [7]. Vogt described 6 types of esophageal atresia:

- Absence of oesophagus,
- Isolated esophageal atresia (Ladd I, Gross A),
- Esophageal atresia with a fistula connecting the proximal portion of the esophagus with the trachea (Ladd II, Cross B),
- Esophageal atresia with a fistula connecting the distal portion of the esophagus with the trachea (Ladd III, Cross C),
- Esophageal atresia with a double fistula connecting both segments of the interrupted esophagus with the trachea (Ladd V, Cross D),
- A tracheoesophageal fistula without esophageal atresia: H or N type fistula (Cross E).

The third variety is the most common and accounts for 85% to 90% of all cases [8].

The motility of the oesophagus is always affected either due to abnormal innervation by an abnormality in neuropeptide distribution [9] or to vagal nerve damage occurring during the surgical repair. The resting pressure in the whole oesophagus is significantly higher than in normal patients and the closing pressure of the lower oesophageal sphincter is reduced.

The trachea is also abnormal in oesophageal atresia. The abnormality consists of an absolute deficiency of tracheal cartilage and an increase in the length of the transverse muscle in the posterior tracheal wall [10].

The diagnosis of oesophageal atresia may be suspected prenatally by the finding of a small or absent fetal stomach bubble on ultrasound scan performed after the 18th week of gestation. Overall the sensitivity of ultrasonography is 42% but in combination with polyhydraminos the positive predictive value is 56% [11]. Polyhydraminos alone is a poor indication of oesophageal atresia (1% incidence).

Available methods of improving the prenatal diagnostic rate include ultrasound examination of the fetal neck to view the blind-ending upper pouch [12] and to observe fetal swallowing and magnetic resonance imaging [13].

In post-natal, the diagnosis is made using A stiff wide-bore (10–12 French gauge) catheter passed through the mouth into the oesophagus. In oesophageal atresia the catheter will not pass beyond 9– 10 cm from the lower alveolar ridge. A plain X-ray of the chest and abdomen will show the tip of the catheter arrested in the superior mediastinum (T2–4) while gas in the stomach and intestine signifies the presence of a distal tracheoesophageal fistula. The absence of gastrointestinal gas is indicative of an isolated atresia. A fine bore catheter may curl up in the upper pouch giving the false impression of an intact oesophagus or rarely it may pass through the trachea and proceed distally into the oesophagus through the fistula. The X-ray may reveal additional anomalies such as a "double bubble" appearance of duodenal atresia, vertebral or rib abnormalities.

There is an increased incidence of associated anomalies in pure atresia (65%) and a lower incidence in H-type fistula (10%). The systems affected are as follows: Cardiovascular 29% (ventricular septal defect and tetralogy of Fallot), anorectal anomalies 14%, genitourinary 14%, gastrointestinal l13% (duodenal atresia, pyloric stenosis and malrotation), vertebral/skeletal 10%, respiratory 6%, and genetic 4%.

Some associations which may include oesophageal atresia are the CHARGE association (coloboma, heart defects, atresia choanal, retarded growth and development, genital hypoplasia and ear deformities), POTTER'S syndrome (renal agenesis, pulmonary hypoplasia, typical dysmorphic facies), SCHISIS association (omphalocoele, cleft lip and/or palate, genital hypoplasia) and VATER association

(vertebral, anorectal, tracheoesophageal and renal or radial abnormalities) that was later expanded as the VACTERL association to include cardiac and limb defects [14].

Chromosomal abnormalities such as trisomies (18 and 21) [15] and deletions (22q11 and 17q22q23.3) [16] are known to be associated with oesophageal atresia and have been reported in up to 6% of patients who have associated malformations in other systems.

Once the diagnosis of oesophageal atresia is established, a suction catheter preferably of the double lumen type, is placed in the upper oesophageal pouch to suction secretions and prevent aspiration occurring during transfer. Definitive management comprises disconnection of the tracheoesophageal fistula, closure of the tracheal defect and primary anastomosis of the oesophagus. Where there is a "long gap" between the ends of the oesophagus, delayed primary repair should be attempted. Only very rarely will an oesophageal replacement be required [17].

Survival is directly related to birth weight and to the presence of a major cardiac defect. In1994, Spitz proposed a new classification of infants born with esophageal atresia into three groups based on birth weight and associated cardiac anomalies [18]:

Group I: Birth weight over 1500 g with no major cardiac anomaly
Group II: Birth weight less than 1500 g or major cardiac anomaly
Group III: Birth weight less than 1500 g plus major cardiac anomaly.

Major cardiac anomaly was defined as either cyanotic congenital heart disease that required palliative or corrective surgery or non-cyanotic heart anomaly that required medical or surgical treatment for cardiac failure.

Infants weighing over 1500 g and having no major cardiac problem should have a near 100% survival, while the presence of one of the risk factors reduces survival to 80% and further to 30–50% in the presence of both risk factors.

Infants with Potter's syndrome (bilateral renal agenesis), trisomy 18 (fatal in the first year of life in over 90% of cases) and with totally uncorrectable major cardiac defects or with Grade IV intraventricular haemorrhage are considered for non-operative management.

CONCLUSION

Esophageal atresia is a congenital failure of the full esophageal lumen to develop. It occurs in 1 in 10,000 births. Careful observation of the fetal abdomen can be helpful. Dilatation of the esophageal pouch can be seen in the upper chest. Over half of affected fetuses have chromosomal abnormalities (mostly trisomy 21) or other morphological abnormalities (especially cardiac).

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