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Case Report

Prenatal diagnosis of esophageal atresia: A case of triple negative screening

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ABSTRACT

Esophageal atresia (EA) is prenatally diagnosed in less than one third of the cases and is usually only suspected. Recently, magnetic resonance imaging (MRI) with dynamic sequence and biochemistry of the amniotic fluid have been proposed to enhance prenatal diagnosis of EA.

We report the case of a triple negative screening (ultrasound, MRI with dynamic sequence and biochemistry of the amniotic fluid) with a postnatal diagnosis of EA type III with a small defect. Even using second line tests, prenatal diagnosis of EA remains a challenge.

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Case report

Esophageal atresia (EA) is a rare congenital malformation with a prevalence of 2.43 cases per 10,000 births [1]. EA is prenatally diagnosed in less than one third of the cases and is usually only suspected [2]. Prenatal suspicion of EA is often based upon the presence of indirect (polyhydramnios and/or the absence of or a small stomach bubble) or direct (pouch sign, deviation of the trachea) signs in ultrasound. Magnetic resonance imaging (MRI) with dynamic sequence and biochemistry of the amniotic fluid have been developed to help in the diagnosis of EA [3]. The combination of ultrasound and of the second line tests could improve antenatal diagnosis of EA [4].

We report the case a 26 years old women with a history of Graves'-Basedow disease (TRAK negative). During the first trimester of her pregnancy, she took Thiamazole switched to Propylthiouracile. The patient was referred to our center and during the third trimester sonography, polyhydramnios was diagnosed associated with an absence of gallbladder. Thyroid was normal and the examination of the esophagus shown no pouch sign and no tracheal print was observed [5]. No other malformation was observed. In this context, amniocentesis at 34 weeks of gestation (WG) was performed and ruled out the diagnosis of cystic fibrosis. Karyotype was also normal. Biochemistry of the amniotic fluid shown a normal EA index (GGTP = 15UI/l and AFP = 0.60 MoM) at 0.5 (normal < 3) [6]. MRI with dynamic sequence at 34 WG, systematically performed in case of prenatal suspicion of EA in our center, concluded to a normal size of the stomach, visualisation of the upper and lower esophagus, no pouch sign and no tracheal bowing (Fig. 1) [7]. Amniodrainage was necessary at 35 WG. The patient was finally induced at 41 WG and delivered vaginally of a girl. EA was diagnosed at birth secondary to the impossibility of the passage of an orogastric catheter. Surgery was performed 24 h after and concluded to an EA type III with a very small defect above the arch of the azygos vein. Anastomosis was done with no tension. The pre operative screening shown no other associated malformation.

This case highlights the difficulty of prenatal diagnosis in EA, even in a referent center and with the combination of ultrasound and second-line tests (MRI with dynamic sequence and biochemistry of the amniotic fluid). Indeed, in case of EA type III, direct visualization of the fluid-filled, blind-ending esophagus during fetal swallowing or upper neck "pouch sign", is more difficult due to absence of stasis. The fistula also leads to having a normal stomach. The small defect does not modify the tracheal print with no tracheal bowing. We can also hypothesis than this absence of stasis does not modify biochemistry of the amniotic fluid.

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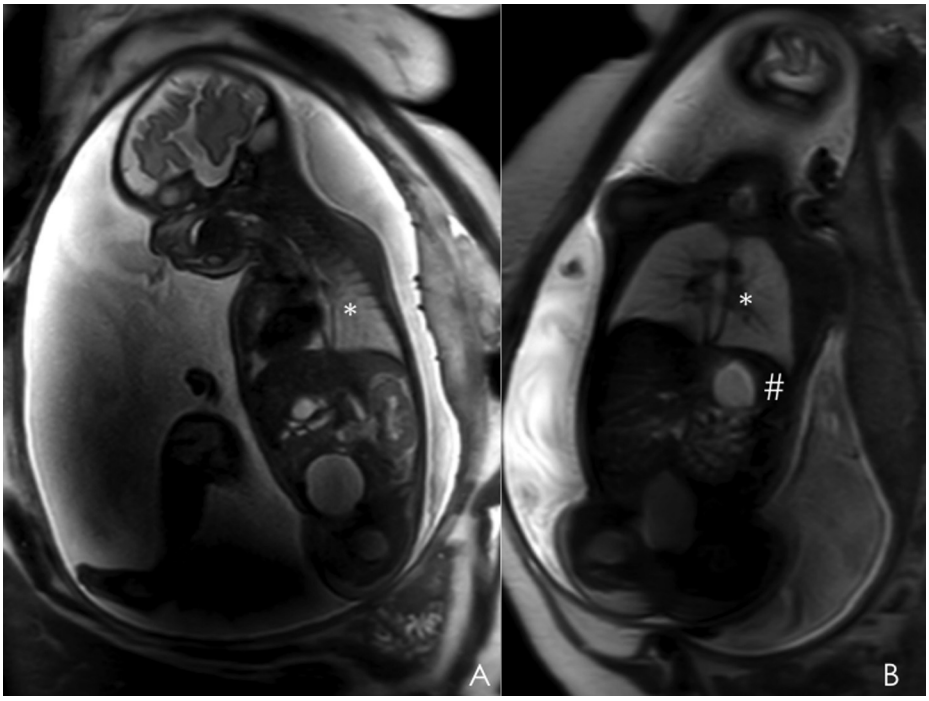

Fig. 1. T2-weighted sagittal (A) and coronal (B) sequence. *: Normal lower esophagus. #: normal stomach.

The prenatal sonographic detection of EA relies on the presence of a small or nonvisible fetal stomach bubble that is associated with polyhydramnios. The positive predictive value of the combination of these 2 signs is appearing to be low and a prenatal sonographic diagnosis can be improved by the direct visualization of the fluid-filled, blind ending esophagus during fetal swallowing or upper neck pouch sign [3]. MRI is interesting in the prenatal diagnosis of EA because it allows for pouch visualization probably more easily than with ultrasound. It avoids some technical pitfalls such as fetal position, but as ultrasound, it requires that the fetus should swallow during the examination. The dynamic sequence helps to visualize the pouch because it captures the moment when the fetus swallows amniotic fluid and should be done in case of EA suspicion [4]. Biochemical analysis consisted on assay of gammaglutamyl transpeptidase (GGTP) and alpha-fetoprotein (AFP) in amniotic fluid. In case of EA, GGTP and AFP are elevated and Czerkiewicz et al defined an EA index (AFP multiplied by GGTP) [6]. Using a cut off of 3 for the EA index, 98% sensitivity and 100% specificity were observed for amniotic fluid prenatal diagnosis of EA, whatever the anatomical type.

In a national cohort, we previously shown than prenatal diagnosis of EA type III was much lower than EA type I (17.9% vs 82.2%, $p < 0.001$), and in particularly in case of small defect [8,9]. Even if prenatal diagnosis does not modify neonatal mortality and outcome at 1-year, it allows antenatal parental counselling, better antenatal management, and avoids postnatal transfers [8,10]. But, in case of EA type III and particularly with small defect, it remains a challenge for next years to improve it.

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