Performance of prenatal diagnosis in esophageal atresia
Obstetrics and maternal-fetal medicine, Hôpital Necker Enfants Malades, AP-HP, Université Paris Descartes, Paris, France

Objective
To investigate the performance of prenatal diagnosis in the detection of esophageal atresia (EA).

Methods
This was a retrospective study from a pediatric surgery database in Necker Hospital about EA management from January 2002 to December 2013. Prenatal data included amniotic fluid volume, stomach visualization and/or size, biochemical amniotic fluid analysis (alpha-fetoprotein (AFP), gamma-glutamyl transpeptidase (GGTP)), magnetic resonance imaging (MRI) results and prenatal diagnosis of associated malformations. Postnatal data included EA type, mortality, and postnatal diagnosis of associated malformations.

Results
122 patients were included. Diagnosis was suspected prenatally in 39/122 (32%) cases. Polyhydramnios was noted in 64/122 (52.4%) cases, stomach bubble was non-visualized or small in 39 (32%). MRI was performed in 28 cases and confirmed EA in 19/28 (68%). Biochemical amniotic fluid analysis was performed in 17 cases and confirmed EA in 15/17 (88%) cases, using a cutoff of 3 for the EA index (AFP multiplied by GGTP). There was 14 (11.5%), 2 (1.6%), 101 (82.8%), 5 (4.1%), and 0 (0%) type I, II, III, IV and V respectively. EA was suspected prenatally in 12/14 (85.7%), 1/2 (50%), 26/101 (25.7%) and 0/5 (0%) in case of type I, II, III, and IV respectively. Associated malformations were detected prenatally in 38 cases and postnatally in 9 cases, therefore a total of 47/122 (39%) cases, including 32 cases of heart defect (26.6%). 19 (15.5%) patients died postnatally, all with type III EA, and 16/19 (84%) had associated malformations.

Conclusion
Prenatal diagnosis of EA remains challenging, especially in type III with no polyhydramnios. Amniotic fluid biochemistry may prove useful in the diagnosis of EA. Prenatal ultrasound examination should also focus on associated anomalies given the high rate of syndromic cases.