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LETTER TO THE EDITOR

Fetal Eagle–Barrett syndrome and pulmonary atresia with intact ventricular septum

Eagle–Barrett syndrome is characterized by the absence of abdominal muscles with urinary tract abnormality and other associated anomalies (OMIM 100100), was first described by Frolich (1839) and presents with a prevalence of about 1/29 000 newborns (Baird and MacDonald 1981). Eagle and Barrett (1950) described 9 cases of congenital deficiency of abdominal musculature with associated genitourinary abnormalities and stated that 42 cases had been reported before their report. Every case except 1 was in a male. They pointed out that genitourinary symptoms are often not present and that the obstructive manifestations in the genitourinary tract must be searched for and corrected at the earliest possible moment. The syndrome is known also with the descriptive appellation of 'prune belly syndrome' because the intestinal pattern is evident through the thin, lax, protruding abdominal wall in the infant. Anatomically, it is caused by urethral obstruction early in development resulting in massive bladder distension and urinary ascites, leading to degeneration of the abdominal wall musculature and failure of testicular descent. The impaired elimination of urine from the bladder leads to oligohydramnios, pulmonary hypoplasia and Potter's facies. The syndrome has a broad spectrum of affected anatomy with different levels of severity and it has been reported to be associated with other anomalies, including persistent urachus, pigeon breast deformity, a variety of gastrointestinal, cardiovascular (such as aortic and pulmonary valve stenosis), urogenital and skeletal abnormalities, and also with pulmonic stenosis, deafness and mental retardation (Baird and MacDonald 1981). It has also been reported to be associated with other genetic conditions, such as Down and VACTERL syndromes (Baird and MacDonald 1981).

Pulmonary atresia with intact ventricular septum (OMIM 265150) accounts for less than 3% of all congenital heart defects with a suggested autosomal dominant inheritance with incomplete penetrance (Grossfeld *et al.* 1997).

A patient was referred to us at 16 gestational weeks of her first pregnancy with a prenatal ultrasonographic diagnosis of massive bladder distension. There was no history of teratological exposures during the first trimester of pregnancy. Fetal chromosomal analysis performed on amniotic fluid sampling showed a karyotype 46,XY. After genetic counselling, the couple decided to perform voluntary termination of the pregnancy. Fetal examination showed a thin, lax, protruding abdominal wall, and autopsy showed complete urethral obstruction and bilateral cryptorchidism and revealed the presence of pulmonary atresia with intact ventricular septum (Fig. 1).

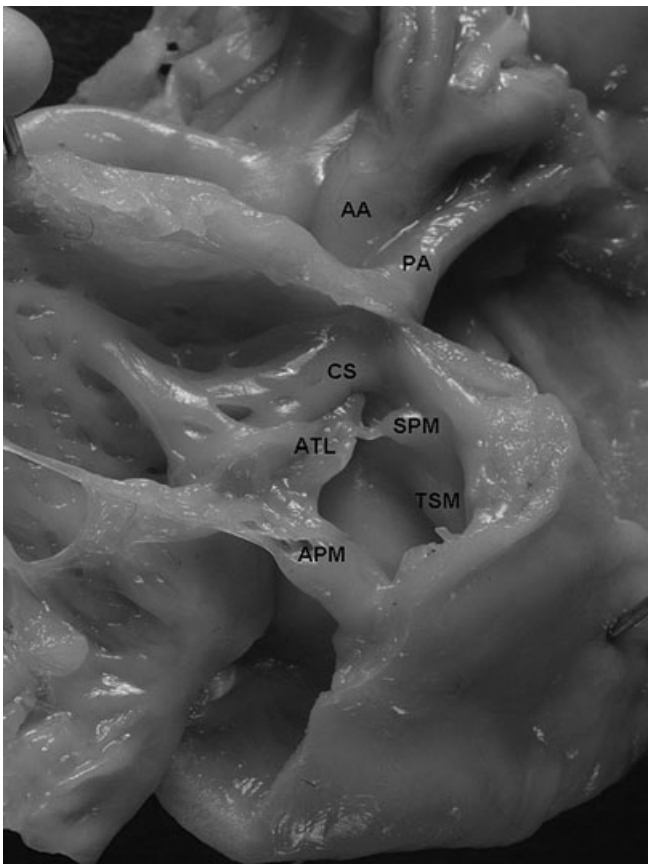

Fig. 1 Gross morphology of right outflow tract; note marked hypoplasia of pulmonary root, atresia of pulmonary valve, marked dilation of the whole ventricular cavity and moderate displasia of tricuspid valve (poor differentiation of cordae with leaflets directly attached to anterior papillary muscle); nevertheless note the tripartite anatomy with inlet, trabecular and outlet components well represented. AA, ascending aorta; PA, pulmonary artery; CS, crista supraventricularis; SPM, septal papillary muscle (Lancisi); ATL, anterior tricuspid leaflet; APM, anterior papillary muscle; TSM, trabecular septo marginalis.

The exact aetiology of Eagle–Barrett syndrome, which occurs primarily but not exclusively in males, is unclear, although several embryologic theories attempt to explain the anomaly. Autosomal recessive inheritance is suggested in some reports. Afifi *et al.* (1972) describe an affected offspring of first-cousin parents born in Lebanon, where the rate of consanguinity is high; Garlinger and Ott (1974) describe two affected brothers in one family and two affected male cousins in a second, and found three other reports of affected relatives, two of affected cousins and one of concordant male twins. Moreover, from the genetic inheritance point of view, if it is an X-linked recessive condition, multiple affected brothers should be observed; however, if the disorder is due to fresh dominant mutation in each case, the male-limitation would be unexpected but not impossible.

Eagle–Barrett syndrome has been reported in association with some congenital heart defects, but an association with pulmonary atresia with intact ventricular septum, to our knowledge, has not been reported in the published literature. There is no embryological connection between the two anomalies, so this association suggests

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that if each of the two conditions is transmitted by autosomal recessive or dominant inheritance, this might be due to contiguous or reciprocally influenced genes.

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