

Unilateral choanal atresia in one monozygotic twin: a case report

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Abstract. – Unilateral atresia is an extremely uncommon congenital disorder in monozygotic twins. We present the first case in the literature of premature, monozygotic twins with no other congenital genetic defects other than only one twin with unilateral choanal atresia on the right side, being the other sibling completely normal. Diagnostic examinations performed to both twins consisted of nasal endoscopy, which revealed only one twin with unilateral choanal atresia; axial computed tomography scan; genetic analysis; chromosome analysis; abdominal ultrasound and echocardiography. In our cases the genetic-chromosomal analysis failed to support a possible cause of unilateral choanal atresia in one monozygotic twin, suggesting a multifactorial etiology.

Key Words:

Choanal atresia, Unilateral, Monozygotic, Twin.

Introduction

Choanal atresia (CA) is a congenital obstruction of the posterior nasal apertures. It affects from one in 5000 to one in 8000 live births, with a female to male predominance of 2:1. CA are classified as unilateral (60%) or bilateral (40%) and bony (90%) with the remaining 10% being membranous. However, some publications suggest that mixed bony-membranous anomalies have the highest incidence^{1,2}. The aim of this paper is to report the first case in the literature of premature, monozygotic twins with no other congenital genetic defects other than only one twin with unilateral CA on the right side, being the other sibling completely normal.

Case Report

In our Department, we had the case of monozygotic twin infant females that were born by cesarean section at 33 weeks of gestation as

third children of healthy non-consanguineous parents. The mother and father were both 39 years old. Pregnancy history has not indicated maternal exposure to smoking, alcohol or illicit drugs. It has to be mentioned that the mother took on the first trimester progesterone capsules 300mg/day per os in 3 divided doses for 3 days, as well as iron, calcium and folic acid supplements. The Apgar scores of Twin A were 5 and 8 and Twin B 5 and 7 at the 1st and 5th minutes, respectively. Twin's A birth weight, height and head circumference were 1550 gm (50th percentile), 34 cm (10 to 50th percentile) and 32 cm (10 to 50th percentile). Twin's B birth weight, height and head circumference were 1450 gm (50th percentile), 32 cm (10 to 50th percentile) and 30 cm (10 to 50th percentile).

Subsequent to delivery, pathologic examination of the placenta revealed a monochorion and diamnion (common placenta and two amniotic cavities). Diagnostic examinations performed to both twins consisted of nasal endoscopy, which revealed only one twin (Twin B) with unilateral CA of bony type; axial computed tomography scan (Figure 1). Genetic analysis, which included chromosome analysis (Figure 2), revealed normal 46, XX female karyotypes without any numerical or structural chromosomal anomaly. Abdominal ultrasound was normal. Echocardiography showed an interatrial communication with a left-to-right shunt (patent *foramen ovale*) in both twins. No other pathological results were observed.

Discussion

Unilateral atresia is an extremely uncommon congenital disorder in monozygotic twins which may remain undetected for years, with unilateral nasal discharge as the only symptom. An upper

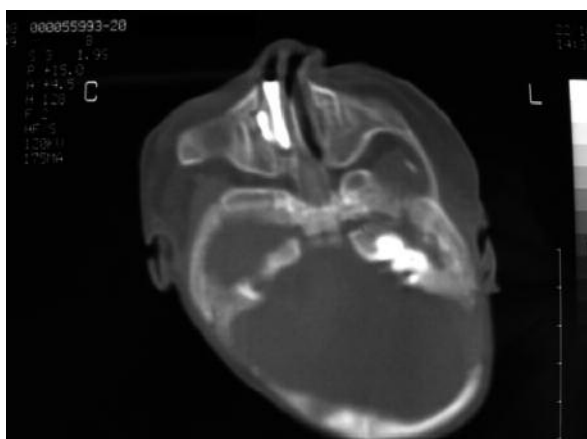


Figure 1. CT imaging of Twin B showing right choanal atresia. Gastrografin remained in the right nasal cavity due to the atresia.

respiratory infection may precipitate symptoms. In unilateral cases, the right side is more commonly affected than the left³.

During the 5th gestational week the nasal pits form on the developing face. The nasal vaults and choanae are completely formed by the end of the 10th week. Consequently, the first 12 weeks of fetal development are the critical time for development of the face. During the first 4 weeks of this period 95% of embryonic formation occurs, when the majority of congenital deformities are initiated¹.

There are different theories to explain the etiology of this disease such as: the persistence

of the buccopharyngeal membrane from the foregut, the persistence of the nasobuccal membrane of Hochstetter, the abnormal persistence or location of mesoderm forming adhesions in the naschoanal region and the misdirection of the neural crest cell migration, and subsequent mesodermal flow^{1,4}.

Non-syndromal CA is usually sporadic and most likely multifactorial. However, there are cases of unilateral CA affecting siblings and affecting successive generations that suggest monogenetic inheritance with autosomal-dominant and autosomal-recessive transmission².

The genetic basis for expression differences (differential allelic expression) in monozygotic twins showed that individual variation is under genetic control. As a result, common allelic variants of genes can be expressed at different levels and thus contribute to phenotypic diversity⁵. In our cases the genetic-chromosomal analysis failed to support a possible cause of unilateral congenital CA in one monozygotic twin, suggesting a multifactorial etiology.

Progesterone induces secretory changes in the endometrium, decreases uterine contractility during pregnancy, and maintains pregnancy. It is not recommended during the first 4 months of gestation. It may cause adverse developmental effects including cardiovascular malformations, hypospadias and female virilization. Other outcomes are miscarriage, preterm birth, stillbirth, neonatal death, low birth weight, fetal genital abnormalities, teratogenic effects and admission to

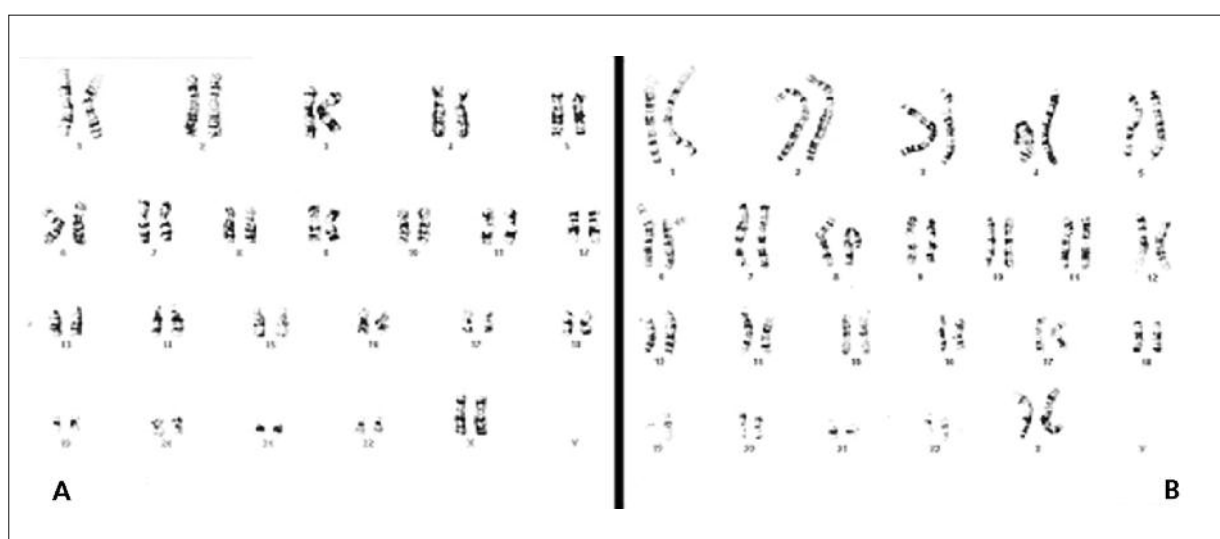


Figure 2. Normal chromosome karyotype (46, XX) of female Twins (**A**) and (**B**). Twin A has no pathological abnormality. Twin B has unilateral choanal atresia.

special care units⁶. In our case the mother took on the first trimester progesterone capsules, which could be a possible etiological factor of congenital CA in twin B.

The most commonly recognized group of associated anomalies is the CHARGE association that includes coloboma of the eye, heart defect, CA, retarded growth and development, genitourinary hypoplasia and ear anomalies and/or deafness. CA has been also associated with Treacher-Collins syndrome and congenital malformations such as syndactyly or polydactyly, microcephaly, esophageal atresia and craniosynostosis^{2,7}.

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