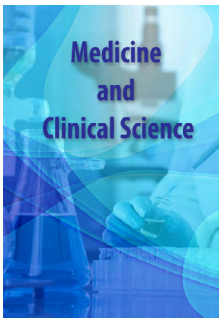
Medicine & Clinical Science

Case Report



Prenatal diagnosis and management of fetal arhinia during pregnancy in order to optimize neonatal outcome

DaCosta Angela, Gittens-Williams Lisa, Williams Shauna and Apuzzio Joseph

Department of Obstetrics, Gynecology & Reproductive Health, Rutgers New Jersey Medical School, Newark, New Jersey, USA

Correspondence

Joseph J Apuzzio MD
Professor & Vice Chair, Director Maternal Fetal Medicine, Department Obstetrics, Gynecology & Reproductive Science, Rutgers New Jersey Medical School, Medical Science Building, E-506, 185 South Orange Ave, Newark, New Jersey 07101, USA
Tel: 973-972-5557
Fax: 973-972-4574
E-mail: Joseph.apuzzio@rutgers.edu

- Received Date: 11 Aug 2022
- Accepted Date: 17 Aug 2022
- Publication Date: 23 Aug 2022

Abstract

Objectives: To discuss the prenatal diagnosis and management of fetal arhinia in order to optimize neonatal outcome

Methods: This is a case report and discussion of the clinical management of fetal arhinia and review of appropriate literature

Results: A fetus with arhinia diagnosed prenatally by ultrasound was managed optimally for neonatal outcome

Conclusion: Prenatal diagnosis of arhinia is a rare congenital malformation that requires consultation and delivery at a specialized children's hospital with the capability of immediate respiratory support at delivery..

Introduction

Arhinia is a partial or congenital absence of the nose structures. It is an extremely rare congenital malformation with an estimate of less than 1,000 total cases in the United States by the National Institutes of Health genetic and rare diseases website (GARD) [1].

Fetal arhinia may be associated with other fetal anatomical structural defects particularly of the upper respiratory tract which if present pose a serious challenge for the newborn at the time of delivery. Due to the rare nature of fetal arhinia, there are no clear guidelines for prenatal diagnosis and management and recommendations of arhinia diagnosed by ultrasound during pregnancy for ancillary providers to be present for newborn respiratory support at the time of delivery. We report a case of prenatal diagnosis of a fetus with arhinia requiring immediate respiratory support at delivery and subsequent tracheostomy in the newborn period.

Case report

The mother is a 27-year-old who was seen in the routine obstetrical clinic of prenatal care. Her history was negative for any fetal or neonatal issues. She underwent a complete obstetric sonogram that was done at 19 weeks gestation. The sonogram was appropriate for gestational age and showed an absence of nasal structures compatible with fetal arhinia (Figures 1 and 2). Other routine maternal screening tests including a serum alfa fetoprotein and a cell-free DNA test were negative.

There was no family history of fetal structural abnormalities. The patient then had a maternal fetal medicine consultation and a specialized sonogram about a week later, which confirmed the same abnormality. It was discussed with the patient the option of amniocentesis with analysis of the amniotic fluid for microarray and karyotype, however the patient declined. A fetal echocardiogram was ordered as well as a consultation with a regional specialized children's hospital was obtained. The fetal echocardiogram did not reveal any structural abnormalities. She was sent for and received a consultation at a regional children's hospital, which confirmed fetal arrhinia. In view of the high-risk for a significant respiratory tract malformation and the need for immediate respiratory tract care at the time of delivery, the patient was transferred for care and delivered at the children's hospital at 36 weeks, 5 days, by cesarean delivery. The indication for the cesarean delivery was a growth restricted fetus with intermittent absent end diastolic flow. A male infant was delivered with an Apgar scores of 2 at 1 minute and Apgar 8 at 5 minutes, birth weight 2,070 kilograms. There was immediate access to the airway chain, which was needed. Physical examination of the newborn was significant for arrhinia, and the remainder of the exam was normal. The infant had a normal karyotype. The newborn required further intervention and subsequently tracheostomy and a G-tube. The plan is for corrective nasal/ respiratory tract surgery at 3 or 4 years of age.

Citation: DaCosta A, Gittens-Williams L, Williams S, Apuzzio J. Prenatal diagnosis and management of fetal arhinia during pregnancy in order to optimize neonatal outcome. Med Clin Sci. 2022; 4(4):1-2

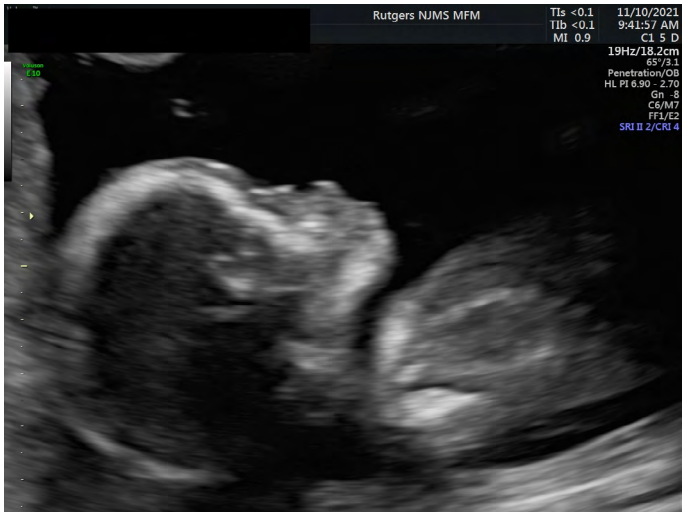


Figure 1. *Two-dimension ultrasound view of fetal profile*

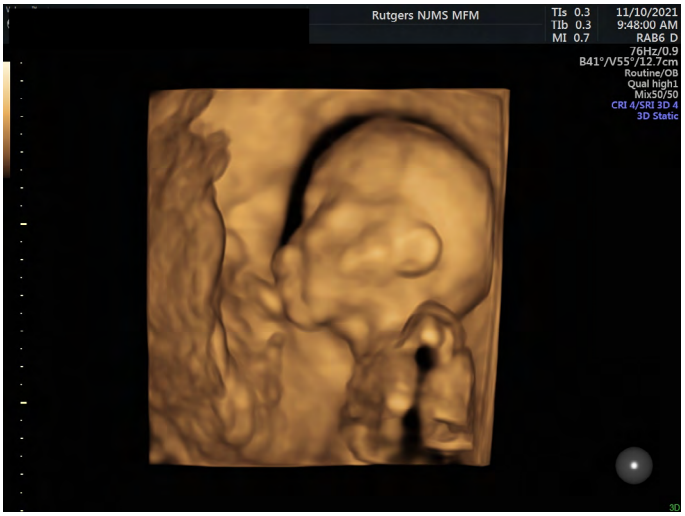


Figure 2. *Three-dimension ultrasound view of fetal profile*

Discussion

Fetal arhinia is a very rare malformation. Most cases described in the literature were diagnosed at the time of delivery and not prenatally [2]. Further management of arhinia when diagnosed during pregnancy should consist of a detailed evaluation of the fetus. This should include a specialized fetal ultrasound examination looking for other fetal structural malformations. A fetal echocardiogram should be performed as well. Other diagnostic studies at the time of diagnosis such as amniocentesis with microarray analysis of the amniotic fluid with microarray should be discussed and offered to the pregnant patient.

The etiology of arhinia is unknown. However, Fuller described a case of arhinia with the neonate found to be heterozygous for the SMCHD1 gene (structural maintenance of chromosomes flexible hinge domain 1) with hypo methylation at the D4Z4 locus. This gene is involved normal development of the nose, eyes and other structures of the head and face [3]. Therefore, it seems prudent to discuss and offer amniocentesis to the pregnant patient with microarray analysis of the amniotic fluid.

Li and colleagues have suggested a fetal MRI to assist in the prenatal diagnosis of arhinia [4]. However, if fetal MRI is not readily available, high-resolution ultrasound of the fetus should be performed and fetal MRI is probably not needed.

In order to optimize the care of the neonate, the pregnant patient was referred to a regional children's hospital that specializes in complex fetal and neonatal diagnosis, treatment and surgery. The hospital is well equipped with case counselors, genetic counselors, Maternal Fetal Medicine specialists, pediatric surgeons, fetal echocardiology specialists and others. The fetus was followed with frequent ultrasound examinations including Doppler studies of the umbilical vessels, fetal weight and biophysical profiles. Fetal growth restriction was diagnosed, and the infant was delivered preterm in a setting where immediate newborn respiratory support was available and needed at delivery.

This case demonstrates the management of fetal arhinia and the significance of this condition since immediately after birth the newborn has respiratory difficulty and required immediate access to the airway team that was present at delivery.

In summary, prenatal diagnosis of fetal arhinia is uncommon. The management plan during pregnancy should consist of a maternal fetal medicine consultation and a specialized sonogram, genetic consultation, maternal screening tests such as cell free DNA testing and genetic studies from the amniotic fluid for microarray and karyotype. These cases require a referral to specialized children's hospital services both during and at the time of delivery in order to optimize the outcome of the newborn.

References

1. https://rarediseases.info.gov/diseases/364/arrhinia
2. Zemtsov G, Swartz A, Kuller J. Prenatal Diagnosis of Arhinia. Amer J of Perin Reports. 2022; 12: 127-130.
3. Fuller A,McCrary H, Graham M,Skirko J. The Case of the Missing Nose. Congenital Arhinia Case presentation and Management Recommendations. Ann Otol Rhinol Laryngol. 2020;19(7): 645-648.
4. Li X, Zhang, Wang F. Prenatal diagnosis of total arhinia by MRI. Jpn J Radiolo. 2015:33(10): 672-674.