ABSTRACT

Background: Arhinia is an embryogenesis defect that leads to the absence of nasal structures and soft tissue (1). It is classified as partial or complete arhinia depending on the presence of the rhinencephalon, which is absent in the complete manifestation and present in the partial manifestation (2).

Only a limited number of congenital arhinia cases have been documented: Less than 100 cases were reported in the literature in the last century (3, 4). However, partial arhinia is still much rarer than complete arhinia (5).

Midline defects or ocular, ear, palatal, or facial abnormalities may co-occur with partial arhinia (6), and it has been associated with somatic anomalies in half of the reported cases (5). The majority of the cases are sporadic, but some familial cases have been reported (7). Arhinia is considered a potentially life-threatening condition that requires neonatal resuscitation by a highly skilled team at the time of delivery (8).

2. OBJECTIVE

In this article, we describe a case of a baby born with congenital partial arhinia at Prince Rashid AlHasan Hospital in Irbid, Jordan.

3. CASE PRESENTATION

A healthy 33-year-old woman (Gravida 3, Para 2) delivered a live female infant by cesarean section at term, after an uneventful pregnancy except for polyhydramnios. During her pregnancy, all of the antenatal ultrasounds were normal. Her social history included a non-consanguineous marriage with a family history free of congenital malformation, and her previous two pregnancies had been normal and had produced one normal boy and one normal girl. However, the current pregnancy was unplanned and the mother had received a Zoladex implant 2 months before the pregnancy for endometriosis. During her pregnancy, all of the antenatal ultrasounds done were normal. Her social history included a non-consanguineous marriage with a family history free of congenital malformation, and her previous two pregnancies had been normal and had produced one normal boy and one normal girl. However, the current pregnancy was unplanned and the mother had received a Zoladex implant for endometriosis 2 months before this pregnancy.

At birth, the baby was dysmorphic, weighed 2.370 kg (7th percentile), was 47 cm long (25th percentile), and had microcephaly, with a head circumference of 29 cm (<1st percentile). The microcephaly was associated with a severely deformed nose with only one stenotic nostril and the presence of a cul-de-sac detected by probing and dilating, in addition to hypotelorism, cleft palate, microphthalmia, micrognathia, and respiratory distress (Figure 1).

The patient was admitted to the neonatal intensive care unit (NICU) and septic workup was done. Four hours later she developed se-
vere respiratory distress, so endotracheal intubation was done. Complete blood count (CBC), blood electrolytes, and blood gases were normal. Chest X-Ray was normal, and an echocardiogram revealed a small atrial septal defect with a patent ductus arteriosus. Both kidneys appeared normal in size and echogenicity by renal ultrasound. The brain ultrasound revealed a mild lateral ventricle dilatation with the absence of the septum pellucidum. Subsequently, a three-dimensional computed tomography scan was done and revealed a single midline mono-ventricle with a dorsal cyst of holoprosencephaly; fused cerebral hemisphere and thalami; and no midline septum pellucidum, corpus callosum, and flax cerebri. She also had a mono-nostril, hypotelorism, and a lobar holoprosencephaly, all features suggestive of partial arhinia. The skeletal survey did not reveal any abnormality, and here karyotype was normal (46 XX). A feeding was started at day 3 of life by orogastric tube and it was tolerated. Two weeks after her NICU admission, the patient developed gram-negative sepsis (*Pseudomonas aeruginosa*) and died due to Disseminated Intravascular Coagulation (DIC).

4. DISCUSSION

Arhinia has a wide clinical spectrum of nose underdevelopment that ranges from partial to complete absence of the nose (9). The disease etiology has not been fully elucidated (5). The likely mechanism suggests defects in medial and lateral nasal process developmental or over development of medial nasal processes and early fusion. Other proposed mechanisms are abnormal migration of neural crest epithelial cells and arrest of nasal epithelial plate absorption from the 13th through the 15th gestational weeks. Nose and nasal cavity development are normally completed during the 3rd and 10th gestational weeks (6).

Less than 100 cases of arhinia have been reported in the literature (3, 4). While there is no significant antenatal history in most of the collected cases (6), in our case the mother had received a Zoladex implant for her endometriosis. However, we did not find any correlation between endometriosis or Zoladex with the incidence of arhinia, although both endometriosis and Zoladex have a risk for congenital abnormalities. Karyotypes in patients with congenital arhinia is mostly normal, the same as in our patient (her karyotype was 46 XX). Familial cases have been reported, but it is believed that most of the cases are sporadic. Translocation between chromosomes 3 and 12 was reported in one case (10), and chromosome 9 aberrations were detected in two cases (11). Clinically, other craniofacial anomalies maybe associated with arhinia including hypo- and hypertelorism, microphthalmia, cleft palate, anophthalmia, coloboma, stenosis of nasolacrimal ducts, absence of paranasal sinuses, ear deformities, optic atrophy, and a wide range of defects in the midline with malformations in the central nervous system, along with mild to severe respiratory distress (1). Our patient had microcephaly, hypotelorism, cleft palate, microphthalmia, and micrognathia. Computed tomography and magnetic resonance imaging should be ordered to detect the possible associated anomalies and to help in surgical treatment planning (6).

The treatment of arhinia focuses mainly on supporting the airway and proper feeding. Ancillary surgical procedures are beneficial to maximize airflow, and if the neonatal respiratory distress is severe a tracheostomy could be done. In our patient, endotracheal intubation was done when she developed severe respiratory distress, about 4 hours after delivery, and an orogastric tube was inserted for feeding on day 3 of life. The plan was to manage her case surgically when she stabilized, but unfortunately she died at the age of 2 weeks due to DIC caused by sepsis. Death before the age of 2 months has been reported in three cases of arhinia. Specifically, one patient died at the age of 2 hours due to respiratory failure and two patients died due to sepsis, one at the age of 29 days and the second at the age of 10 weeks (12). The limitation of this article is that a genetic study was not completed for this patient.

5. CONCLUSION

Congenital partial arhinia is a developmental defect with an unknown etiology and an extremely rare incidence. It could be associated with dangerous life-threatening complications such as feeding and airway compromise. Hence, management of the condition is essential and urgent, including surgical correction as needed by a highly skilled team.

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• Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms.

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