Case Report

Unilateral Pulmonary Agenesis with Mayer-Rokitansky-Kuster-Hauser Syndrome: A Rare Association

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Abstract

Unilateral pulmonary agenesis is a rare congenital condition characterized by the absence of one or both lungs. Most of the cases have unilateral presentation as bilateral agenesis does not support life. Simultaneous Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome also has very rare presentation. It is characterized by congenital aplasia of the uterus and the vagina with normal female karyotype (46XX). A 30-year-old female, presenting with dyspnea from last 6 months and amenorrhea for last 15 years was further evaluated with gynecological examination, contrast-enhanced computed tomography thorax with the abdomen and karyotyping. She was eventually diagnosed as a case of Type I pulmonary agenesis with Type I MRKH syndrome. K

Keywords: Contrast-enhanced computed tomography thorax and abdomen, Mayer-Rokitansky-Kuster-Hauser syndrome, unilateral pulmonary agenesis

Introduction

Pulmonary agenesis is a rare disorder which occurs congenitally. Most of the cases have unilateral presentation as bilateral agenesis does not support life.[1] In 1673, Depozze noted the first case of unilateral pulmonary agenesis while doing autopsy of a female body.[2] The reported incidence is 1 in 15,000 births.[3] In around 50% cases with unilateral pulmonary agenesis, it accompanies anomalies of other organ system such as central nervous system, cardiovascular system, gastrointestinal system, genitourinary system, skeletal system, and also rarely associated with Klippel–Fiel syndrome and Down syndrome.[4] The clinical presentation may range from asymptomatic individual to individuals with complaint of recurrent chest infections, shortness of breath, and severe respiratory distress. The spectrum of onset of symptoms may range from early neonatal period to childhood and even during adult life.[4]

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a very rare disorder characterized by congenital aplasia of the uterus and the vagina with normal female karyotype (46XX). The incidence of MRKH syndrome is around 1 in 4500 female births.[5] Primary amenorrhea is the main presenting symptom with normal genitalia and normal secondary sexual characteristics. MRKH can also be associated with renal, skeletal, hearing, and cardiac anomalies. Unilateral pulmonary agenesis is rarely associated with this syndrome with fewer cases have been reported in the literature. We are reporting an interesting case who presented with main symptom of shortness of breath and amenorrhea and after evaluation diagnosed as a case of unilateral pulmonary agenesis with MRKH syndrome.

Case Report

A 30-year-old female presented to our outpatient department with complaint of shortness of breath for last 6 months which was gradual in onset and slowly progressive. She had no other

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respiratory complaints. She had no past history of tuberculosis. On further inquiry, she had history of amenorrhea for last 15 years. With the above complaints, she was advised a chest X-ray, routine blood investigations, and gynecologist opinion for complaint of amenorrhea. The chest X-ray showed right middle and lower zone homogenous opacity with obliteration of the right costophrenic and cardiophrenic angle with mediastinal shift toward right [Figure 1]. On auscultation, breath sounds were absent on right side, and on gynecological examination, her vaginal passage was 1 cm long and had a blind end. Her ultrasonography thorax revealed no collection in the right pleural cavity but showed the presence of the heart in the right hemithorax. The patient was then advised for contrast-enhanced computed tomography (CECT) thorax with the abdomen and karyotyping for ruling out congenital anomaly. The CECT thorax and whole abdomen revealed complete absence of the right lung with hyperinflation and herniation of the opposite lung into the right thoracic cavity with the absence of uterine cavity in the pelvis [Figure 2a and b]. The karyotyping came out to be normal, i.e., 46XX. From above findings, our patient was diagnosed as a case of Type I pulmonary aplasia with Type I MRKH syndrome.

Discussion

Unilateral pulmonary agenesis is a rare congenital anomaly with reported incidence of 1 in 15,000 births. Owing to its common association with other organ malformations, only a few cases live to adult life. This patient had presented at the age of 30 years with respiratory symptoms from last 6 months. Boyden modified the classification given by Schneider and Schwalbe and divided the pulmonary agenesis into three types: Type 1 known as pulmonary agenesis in which the bronchus and the lung are absent with the absence of its blood supply. Type 2 known as pulmonary aplasia in which pulmonary parenchyma is absent with the presence of rudimentary bronchus. Type 3 known as pulmonary hypoplasia in which there is underdevelopment of the lung parenchyma, bronchial tree, and its blood supply.[6]

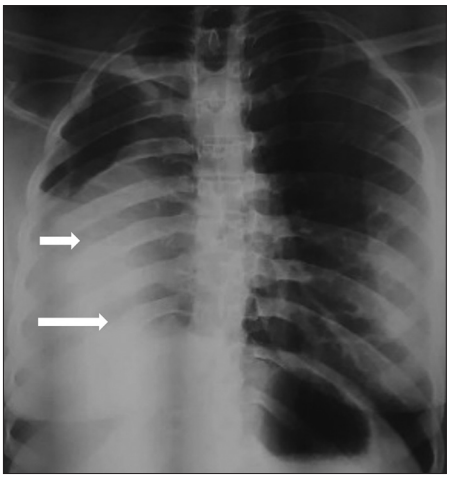

Figure 1: Chest X-ray showing right middle and lower zone homogenous opacity with obliteration of the right costophrenic and cardio phrenic angle with mediastinal shift toward right

MRKH syndrome is the second most common cause of primary amenorrhea after gonadal dysgenesis and it is characterized by aplasia of the upper two-thirds of the vagina and the uterus along with normal ovaries, fallopian tubes, secondary sexual characters, and 46XX karyotype. It is caused due to the interrupted development of the Mullerian duct system, which normally forms the uterus, cervix, and upper 2/3rd of vagina, during 5th and 6th week of gestation.[7]

The cases of MRKH syndrome are infrequently reported with unclear etiology. There is an autosomal dominant mode of inheritance with variable expression and incomplete penetrance.[8] MRKH syndrome on the basic of anatomical feature is divided into two types: Type I is isolated uterovaginal aplasia also known as Rokitansky sequence and it is less common in presentation. Type II is more common in presentation and is also known as MURCS (Mullerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia). There is partial aplasia associated with other organ malformations such as skeletal which includes Klippel–Feil anomaly or fused vertebrae (mainly cervical) and scoliosis. Renal involvement may present as unilateral agenesis, ectopia of the kidneys, or horseshoe kidney. It can also accompany cardiac, digital anomalies (syndactyly and polydactyly), and hearing defects. It is also known as genital renal syndrome or MURCS association. During gastrulation, if there is disruption in the migration of mesoderm, it can lead various abnormalities. If paraxial mesoderm is involved, it leads to defects in the development of cervical vertebrae. Involvement of intermediate mesoderm will lead to defects in development of urogenital structures. Limb defects occur if there is disruption in the growth of lateral plate mesoderm.

A case reported by Bachh *et al*. showed the presence of MRKH with agenesis of the right lung along with rudimentary right bronchus, right renal agenesis with ectopic pelvic left kidney.[9] In our case, there was complete absence of the right main bronchus with the absence of the uterus and upper part of the vagina with no other organ system involvement. Another case reported by Shivalingappa and Shetty showed

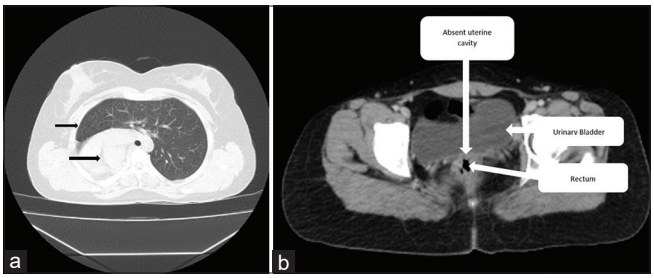

Figure 2: (a)Contrast-enhanced computed tomography chest showing complete absence of the right lung with hyperinflation and herniation of the opposite lung into the right thoracic cavity (b) contrast-enhanced computed tomography abdomen showing the absence of uterine cavity in the pelvic cavity

the presence of Type II MRKH with Type II pulmonary agenesis.[10] Our case was classified as Type I pulmonary agenesis with Type I MRKH syndrome, which is a rare association.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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