



## CASE REPORTS

# Unilateral Agenesis of Diaphragm with Bilateral Renal Agenesis: An Anomaly Diagnosed Prenatally During a NT Scan (11–14 Weeks)

N. S. Shashi Kumar<sup>1</sup> Received: 8 December 2021 / Accepted: 1 May 2022 / Published online: 16 August 2022  
© Society of Fetal Medicine 2022

**Abstract** Unilateral agenesis of the diaphragm with bilateral renal agenesis is an extremely rare lethal developmental anomaly. A detailed evaluation of fetal organs during nuchal translucency scan is required to diagnose this type of developmental anomaly. Detecting lethal developmental abnormality at an early gestational period (11–14 weeks) will be invaluable in facilitating the decision to terminate the pregnancy. Termination in the first trimester reduces psychological stress to the parents. We present a rare case of combined developmental anomaly which has never been reported in the literature.

**Keywords** Nuchal translucency · Transabdominal scan · Transvaginal scan · Agenesis of the diaphragm · Bilateral renal agenesis

## Introduction

Congenital diaphragmatic agenesis is a rare type of diaphragmatic defect with an incidence of 1:250,000 births [1]. It can be unilateral or bilateral, and it is characterized by the herniation of abdominal viscera into the thorax [1].

## Case Report

A routine antenatal check-up and nuchal translucency scan were done on a 23 year old primigravida in her 3rd month

of pregnancy after spontaneous conception. There were no antenatal risks for structural abnormalities like consanguinity or drug intake.

Transabdominal scan was performed using a GE Voluson P8 unit. The nasal bone was seen. The nuchal translucency had a normorange thickness. The heart was seen pushed to the right hemithorax and the stomach was next to it. Transvaginal sonography (TVS) was subsequently done for a better understanding of the structural defects. Herniated contents were seen more clearly.

At 14 weeks, a prenatal ultrasound revealed a single live fetus, (Figs. 1, 2, 3) with herniation of the stomach, small bowel and left lobe of the liver into the left hemithorax. Malrotation of the heart towards the right side was noted. Both lungs and mediastinum were pushed towards the right side. Both kidneys were not seen. Amniotic fluid appeared normal. Growth was corresponding to the gestational period.

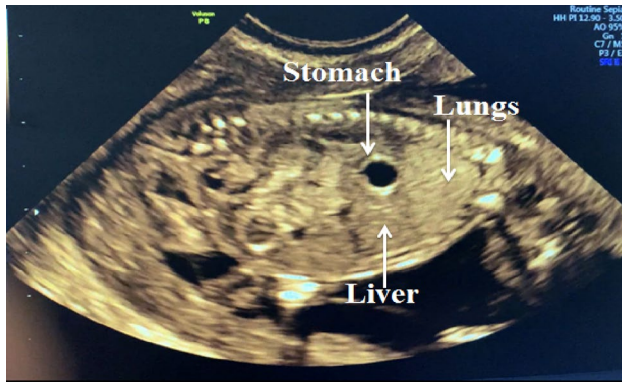
The couple was counseled about this lethal anomaly.

As it was an extremely rare condition, the chance of recurrence is very minimal. The mother was advised a detailed first trimester evaluation in all future pregnancies.

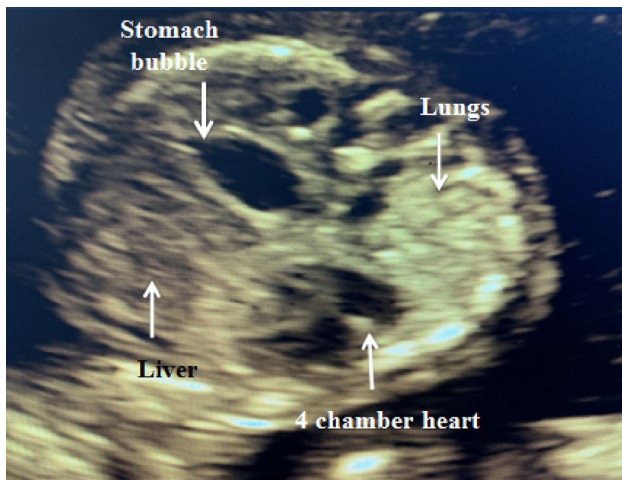
The couple opted for termination of pregnancy. Subsequently, autopsy and microarray were done. Microarray was normal.

✉ N. S. Shashi Kumar  
drshashikumarns@gmail.com

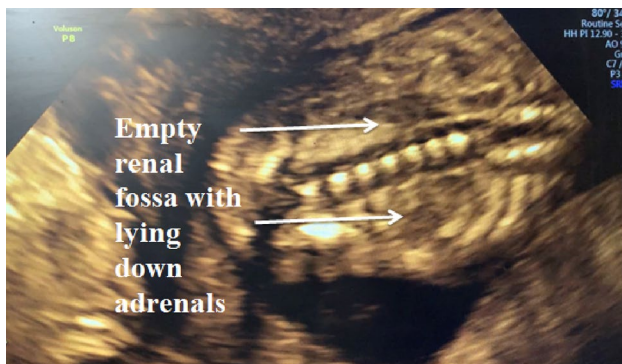
<sup>1</sup> Subbaiah Institute of Medical Science, Shimoga, Karnataka, India



**Fig. 1** Sagittal section showing Stomach bubble, liver, and left lung side by side in left hemithorax



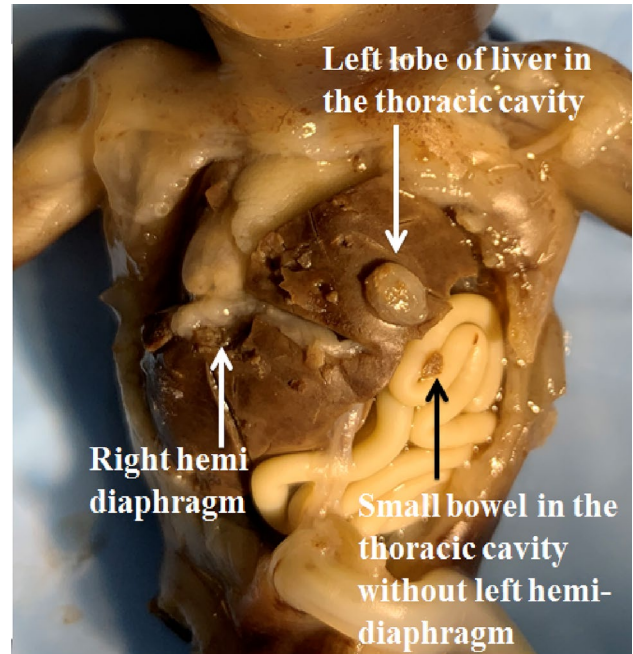
**Fig. 2** In the 4 chamber view stomach bubble, liver and lung seen along with the heart



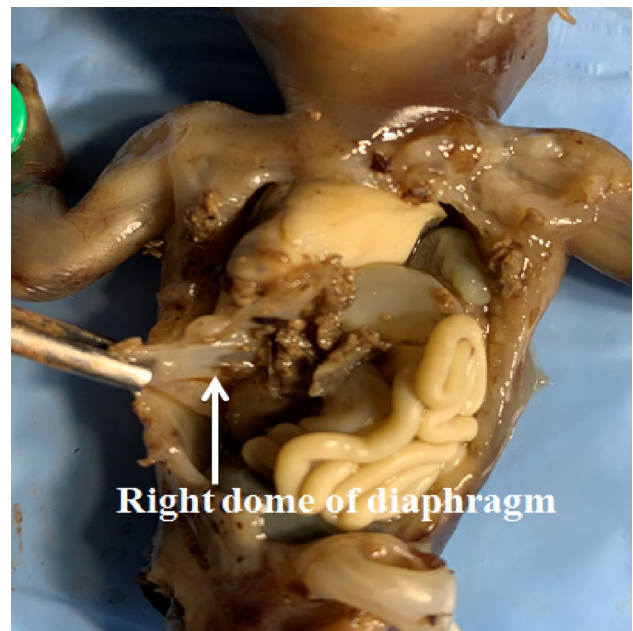
**Fig. 3** Bilateral non visualisation of the kidneys with bilateral "lying down" adrenals

**Autopsy of the Fetus**

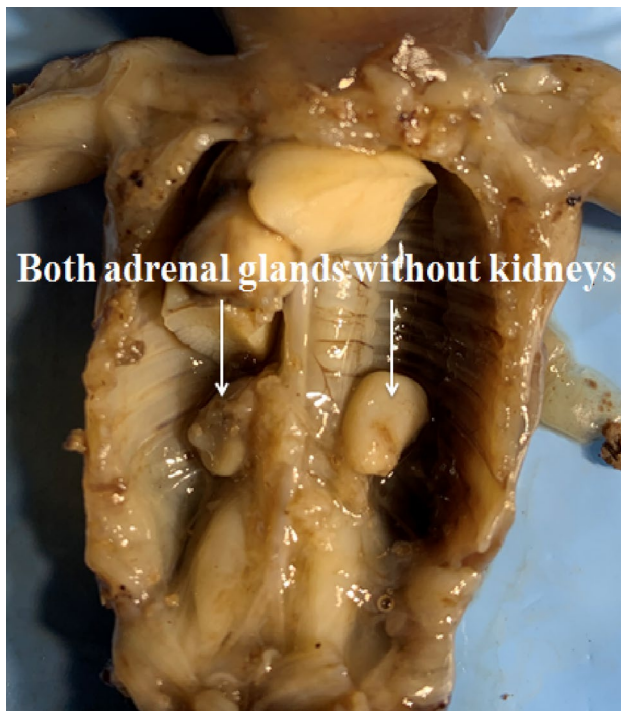
See Figures 4, 5 and 6



**Fig. 4** Left hemithorax filled with stomach, small bowel loops and left lobe of the liver. Mediastinum pushed towards the right. Intact right hemidiaphragm. Left hemidiaphragm not seen. Slight rotation of the heart towards the right



**Fig. 5** Right dome of diaphragm was normally developed. Complete agenesis of the left dome of the diaphragm. Both lungs were hypoplastic and pushed towards the right hemithorax



**Fig. 6** Bilateral renal agenesis. Absent gall bladder

## Discussion

Development of the diaphragm begins in the third week of pregnancy from four primordia: Septum transversum, pleuroperitoneal folds, mesentery of the esophagus, and cervical somite myotomes. Septum transversum is positioned in the intraembryonic cavity and divides it into the pleuropericardial cavity and the peritoneal cavity, maintaining communication through the pleuropericardial canals [2]. This allows herniation of visceral content into the thoracic cavity and interferes with normal lung development. The severity of these adverse effects varies depending on the gestational age at the time of herniation of visceral organs.

Agenesis of the diaphragm results from the failure of full development of one of these four elements. Congenital diaphragmatic defects can be sporadic or a component of various syndromes, some of them with chromosomal anomalies [3]. The presence of the liver in the thoracic cavity makes the prognosis poor [4]. The abdominal contents herniate into the thoracic cavity and compress the lungs and often cause a shift of the mediastinum. The herniation coincides with the critical period of lung development when bronchial branching and pulmonary artery development occurs. The consequence is a marked reduction in terminal bronchioles, thickened alveolar septae, reduced alveolar volume and increased arterial medial wall thickness. This results in fixed increased vascular resistance and reduced surface area for gas exchange with resultant lung compression causing

pulmonary hypoplasia. Postnatal compromise is the result of the fixed pulmonary and vascular hypoplasia, and reversible, pulmonary vascular reactivity. This results in respiratory failure and persistent fetal circulation. These features are the main determinants of neonatal survival.

An autopsy revealed (Figs. 4, 5 and 6) herniation of the stomach, small bowel loops and left lobe of liver into the left hemithorax. Malrotation of the heart towards the right side was noted. Both lungs and mediastinum were pushed towards the right side. Both kidneys were not seen. Surprisingly, there was complete agenesis of the left dome of the diaphragm as against the diaphragmatic defect opined in the prenatal scan. After the autopsy, the final diagnosis was unilateral agenesis of the diaphragm with bilateral renal agenesis. Diaphragmatic defects are often seen. Unilateral agenesis of the diaphragm is extremely rare and limited data is available in the literature. This combination of unilateral agenesis of the diaphragm with bilateral renal agenesis has never been reported. Apart from all the above findings the gall bladder was absent too [5].

## Conclusion

Over 60% of CDH cases are initially suspected on a routine sonographic fetal anatomic survey at 18 to 22 weeks of gestation [6]. The NT scan should be considered as an early anomaly scan. An attempt must be made to look for any structural abnormalities. Whenever more than one system is involved, we recommend DNA storage, to rule out single gene disorders in the future, if needed.

## Learning Points from this Case Report

1. The NT scan should be considered an early anomaly scan. An attempt must be made to look for any major structural abnormalities.
2. Judicious use of transvaginal approach is invaluable in identifying various structural abnormalities.
3. Had this not been diagnosed in the first trimester, anhydramnios in the second trimester would have made the antenatal diagnosis of a diaphragmatic defect difficult.

**Acknowledgements** I acknowledge consultant obstetrician and gynaecologists Dr. Nirmala H. T, Dr. Pallavi K. V and Dr. Sameena Sulthana for their valuable inputs in counselling and managing the case.

**Funding** No funding received.

## Declarations

**Conflict of interest** The case report has been published as a poster in 29th Annual, POGS conference REEDIFY 2021, 6-7th march 2021 at PUNE conducted by Pune Obstetric and Gynaecological Society. The author declares that there is no other conflict of interest.

**Ethical Approval** Since this is a Case Report, no ethical approval was required.

**Informed Consent** Written informed consent was obtained from the parents for publishing the data, publishing images and for autopsy.

## References

1. Morosanu C, Cioca A, Stamatian F, Crisan D. Prenatal diagnosis of unilateral agenesis of diaphragm and associated anomalies. *Indian J Pathol Microbiol.* 2016;59(02):251–2.
2. Wells LJ. Development of the human diaphragm and pleural sacs. *Contrib Embryol.* 1954;35:107–34.
3. Roy M, Chikkannaiah P, Bali A. Congenital diaphragmatic hernia with hypoplastic lungs, heart, and additional anomalies: A case of? Fryns syndrome. *J Dr NTR Univ Health Sci.* 2013;2:130–2.
4. Paek BW, Danzer E, Machin GA, Coakley F, Albanese CT, Filly RA. Prenatal diagnosis of bilateral diaphragmatic hernia: diagnostic pitfalls. *J Ultrasound Med.* 2000;19:495–500.
5. Marlow J, Thomas J. A review of congenital diaphragmatic hernia. *Australas J Ultrasound Med.* 2013;16(1):16–21.
6. Ishak A, Abu Hassan H, Mohd Jamil AA, et al. Role of predictive mortality calculator in antenatal assessment of congenital diaphragmatic hernia. *J Fetal Med.* 2021;8:211–5.

**Publisher's Note** Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.