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CASE REPORT Open Access

# Heterotaxy syndrome with mixed bronchopulmonary malinosculation: an unusual case report

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## Background

Heterotaxy, in simple terminology, can be described as abnormal orientation of abdominal and thoracic organs to the left and right of the central axis of the body, along with presence of a wide variety of complex congenital cardiac as well as extracardiac lesions [1]. The patients can be classified into either right or left isomerism depending upon the morphology of the paired structures which are mirror images present bilaterally in an otherwise asymmetric anatomy [2].

While there have been case reports on heterotaxy, very few of them have documented an association with lung parenchymal anomalies or pulmonary malinosculation. Pulmonary malinosculations can have bronchial, arterial and venous components either individually, or in combination. To the best of our knowledge, there are only two case reports showing association of heterotaxy syndrome with lung anomalies one of which mentions polysplenia with congenital lobar emphysema while the other describes polysplenia with diffuse pulmonary arteriovenous malformations [3, 4]. This is the first report highlighting association of heterotaxy syndrome with mixed bronchoarteriovenous pulmonary malinosculation and the combination of imaging features eventually altered the treatment protocol for the patient significantly.

## Case description

A 10-year-old female child presented to the outpatient department with chest pain and dyspnea for two years which had gradually progressed over time. She had no prior hospital admissions or any significant family history. She underwent echocardiography which revealed atrial septal defect (ASD) with features of right-sided volume overload. A cardiac computed tomography (CT) scan was subsequently planned for detailed structural evaluation and surgical planning.

CT scan demonstrated bilateral hyparterial bronchi, polysplenia, midline liver with preduodenal portal vein (Fig. 1A, B) and reversed SMA-SMV relation. There was superior sinus venosus ASD with right-sided partial anomalous pulmonary venous connection (PAPVC) into the right atrium-superior vena cava (SVC) junction (Fig. 2A, B). All these features were consistent with heterotaxy syndrome. Other notable findings were common origin of left common carotid and innominate artery and dilated main pulmonary artery suggesting pulmonary arterial hypertension.

Lung window showed an unusual bronchopulmonary lung anomaly where right lower lobe contained a well-defined hypoattenuating lesion with multiple intralesional cystic areas of ~ 0.5 to 2 cm (Fig. 3A). It showed no communication with parent tracheobronchial tree except for the presence of a rudimentary isolated bronchus at its medial aspect. On reviewing CT for vascular supply, lesion was seen to derive arterial supply from descending aorta via multiple channels (Fig. 3B). However, the venous drainage was into right inferior pulmonary vein (PV) which further showed anomalous

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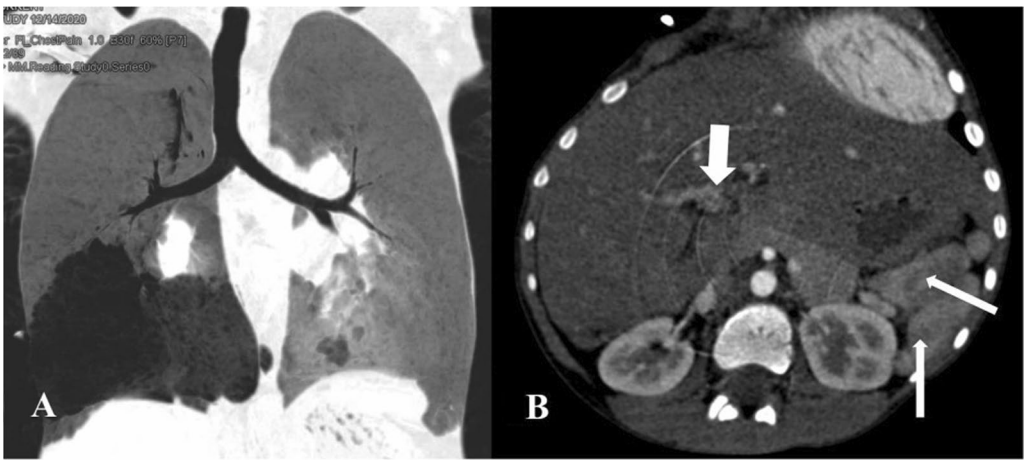


**Fig. 1** Contrast CT images in lung (**A**) and mediastinal (**B**) windows show bilateral hyparterial bronchi with midline transverse liver, polysplenia (thin arrows) and preduodenal portal vein (thick arrow)

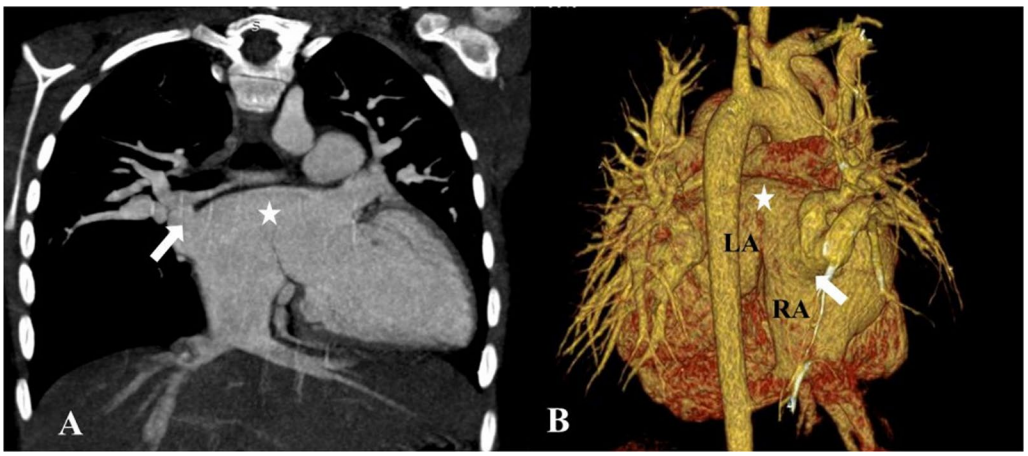


**Fig. 2** **A**, **B** Coronal MPR and VRT images show right-sided partial anomalous pulmonary venous drainage (arrow) with atrial septal defect (*)

drainage into right atrium-SVC via a common channel formed with right superior PV. No separate pleural covering or consolidation was noted.

Following discovery of this anomaly, a complete diagnosis of heterotaxy syndrome with Type G bronchopulmonary malinosculation was made which changed the surgical plan from pericardial patch closure of ASD and baffling of right PV into left atrium to an added right lower lobectomy. No procedural complications were encountered and the patient had an uneventful recovery.

Histopathology of the resected lung lobe showed variable sized cystically dilated spaces lined by bronchial epithelium (Fig. 4,5) and was consistent with hybrid lesion showing intralobar sequestration and Type II congenital pulmonary airway malformation (CPAM).

## Discussion

Disordered determination of left–right axis of body during the embryological development results in heterotaxy [5]. The abnormal orientation of thoraco-abdominal organs can lead to situs ambiguus where there is visceral malposition and dysmorphism associated with indeterminate atrial arrangement [6]. Isomerism in context of congenitally malformed heart is defined as a situation where some paired structures on opposite sides of the left–right axis of the body are, in morphological terms, symmetrical mirror images of each other [1]. An easy approach to decide left or right isomerism can be followed by determining the bronchial situs, atrial morphology and presence or absence of spleen (Table 1). Patients with right isomerism almost invariably present with complex cardiac anomalies such as a single ventricle, large

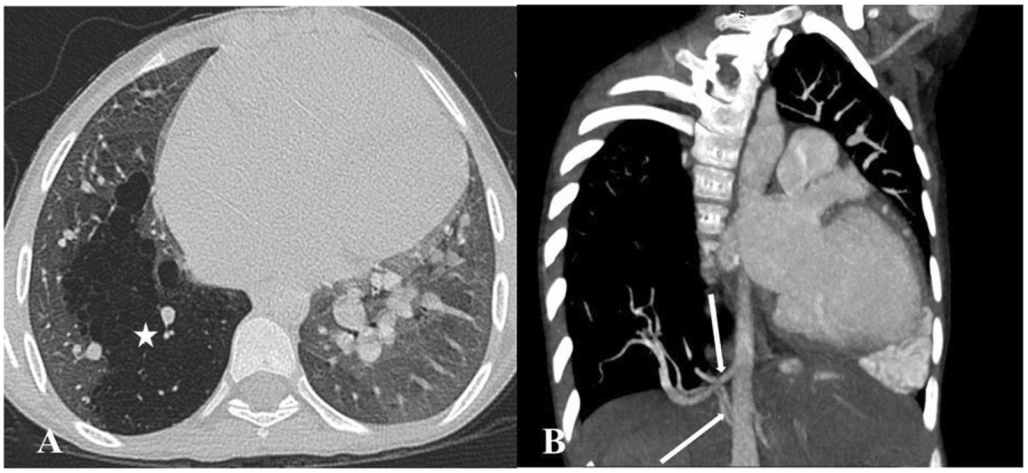


**Fig. 3** **A**, **B** Axial lung and coronal oblique mediastinal window showing hypoattenuating lung lesion (*) with variable sized cystic areas showing anomalous systemic arterial supply from abdominal aorta (arrows)

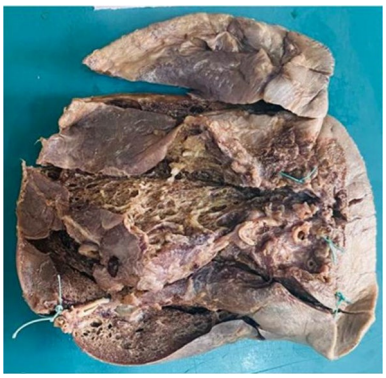

**Fig. 4** Resected gross specimen of right lower lobe showing multiple small cystic spaces varying in size from 0.2 to 1 cm in center as well as the periphery with no visible areas of hemorrhage, calcification or mass

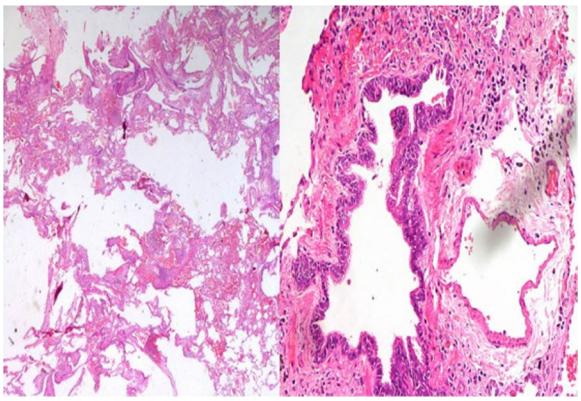

**Fig. 5** Multiple sections identified from the cystic area in the lung on hematoxylin and eosin stain show presence of multiple variable sized cystically dilated spaces lined by bronchial epithelium. Along with it, many dilated ectatic alveolar spaces area seen

ventricular septal defect, pulmonic stenosis or atresia resulting in cyanosis which is the commonest presenting feature. Clinical findings in patients with left isomerism are non-specific with relatively less severe cardiac defects which include endocardial cushion defects, double outlet right ventricle or PAPVC [2]. Extracardiac abnormalities are commonly seen, with a high incidence of anomalies of renal tract, biliary system as well as intestinal malrotation. State of spleen must also be assessed in all cases of isomerism. Asplenia in right isomerism predisposes individuals to lifelong risk of fulminating infections [1].

Associated lung anomalies are rarely seen in cases of heterotaxy. Limited case reports are found in literature. Choh et al. [3] reported one case of left isomerism with congenital lobar emphysema while Kapur et al. [4]

**Table 1** Methodological approach for determination of type of isomerism

| |
|---|
| Step 1: Look at the bronchial situs |
| Bilateral Eparterial bronchi |
| Bilateral hyparterial bronchi |
| Step 2: Look at the bi-atrial morphology (on the basis of appendage which is broad in right atrium and narrow/tubular in leftatrium) |
| Step 3: Look at the splenic status |
| Asplenia |
| Polysplenia |

Bilateral eparterial bronchi with bilateral right atria and asplenia—Right isomerism

Bilatral hyparterial bronchi with bilateral left atria and polysplenia—Left isomerism

**Table 2** Classification of bronchopulmonary malinosculation by Lee et al

| Types | Anomaly | Examples |
|---|---|---|
| A | Isolated bronchial PM | Tracheal stenosis, tracheal bronchus |
| B | Isolated arterial PM | Interrupted pulmonary artery |
| C | Isolated venous PM | PAPVC, TAPVC |
| D | Mixed bronchoarterial PM | Typical intralobar sequestration |
| E | Mixed bronchovenous PM | Combination of type A and C |
| F | Mixed arteriovenous PM | Fistula between systemic pulmonary artery and vein |
| G | Mixed bronchoarteriovenous PM | Classic scimitar syndrome, extralobar sequestration |

reported another case of left isomerism with pulmonary arteriovenous malformations. Congenital bronchopulmonary vascular malformations encompass a wide spectrum of disorders with abnormality of either the components of lung parenchyma/airway, arterial supply or venous drainage. Communication between the three different components is referred to as malinosculation [7]. Lee et al. [8], classified bronchopulmonary malinosculation into seven types (Table 2) which in our case fit into mixed bronchoarteriovenous pulmonary malinosculation (Type G) due to presence of sequestration and CPAM in addition to PAPVC.

The multiplicity and diversity of findings involving various systems in such cases makes individualization extremely valuable, since most of them do not perfectly fit into any one category of the classification. The role of radiological evaluation thus becomes indispensable for correct identification as well as appropriate surgical planning of the patient.

## Conclusion

Heterotaxy syndrome with lung parenchymal anomaly is an extremely rare condition which requires detailed anatomical reporting to help decide management and plan surgical procedures. Imaging studies especially CT scan play a crucial role in identifying such lesions preoperatively and therefore should be appropriately utilized in cases of congenital cardiac defects. A combination of heterotaxy with mixed bronchoarteriovenous pulmonary malinosculation has not been previously described and needs to be actively ruled out as failure to recognize these anomalies may lead to redo surgeries in the future.

#### Abbreviations

ASD: Atrial septal defect; CPAM: Congenital pulmonary airway malformation; CT: Computed tomography; PAPVC: Partial anomalous pulmonary venous connection; PV: Pulmonary vein; SMA: Superior mesenteric artery; SMV: Superior mesenteric vein; SVC: Superior vena cava.

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#### Authors' contributions

VG involved in substantial contributions to the conception provided, design of the work, interpretation of the data and manuscript draft. PG involved in substantial contributions to the conception provided, design of the work, interpretation of the data and substantial revision of the manuscript. KB involved in substantial contributions to analysis of the data and interpretation and revision of the manuscript. NA and SP involved in substantial contributions to the conception provided and analysis of the data and interpretation. PN and RK involved in substantial contributions to the analysis of the data and interpretation. All authors read and approved the final manuscript.

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#### Availability of data and materials

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

## Declarations

#### Ethics approval and consent to participate

The patient included in this study provided the written informed consent to participate in this research.

#### Consent for publication

The patient included in this research gave a written and informed consent to publish the data contained within this study.

#### Competing interests

The authors declare that they have no competing interests.

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