KREP

2025

KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE PRESENTATION

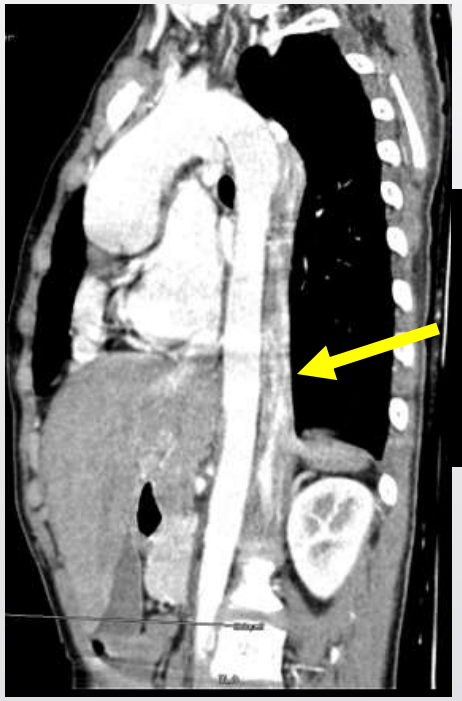
CASE OF HETEROTAXY SYNDROME (CARDIAC ISOMERISM)

MENTOR: DR SANTOSH PATIL
KAHER UNIVERSITY
J.N.MEDICAL COLLEGE ,BELAGAVI
PRESENTER: LIPI AGARWAL

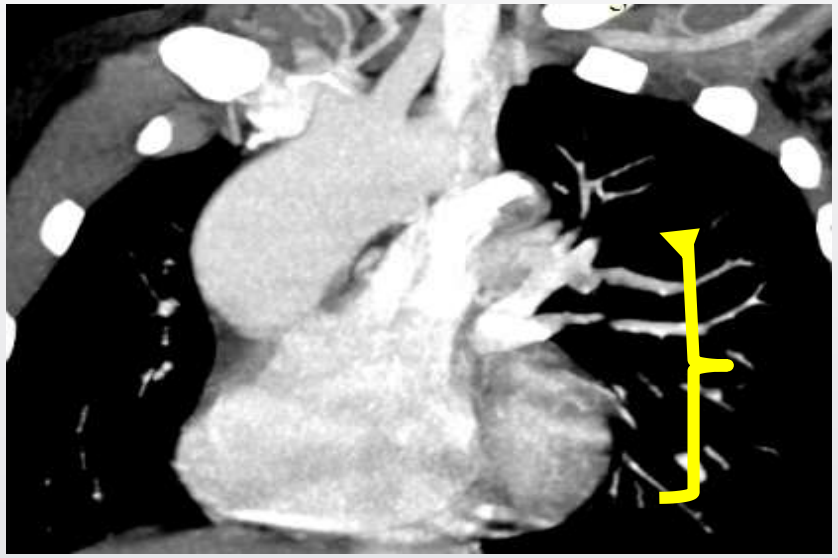
Case 1

- CASE 1 -12YRS/ F- presented with history of shortness of breath and cough
- Presented to the KLE's radiology department for further evaluation and underwent CT pulmonary angiogram

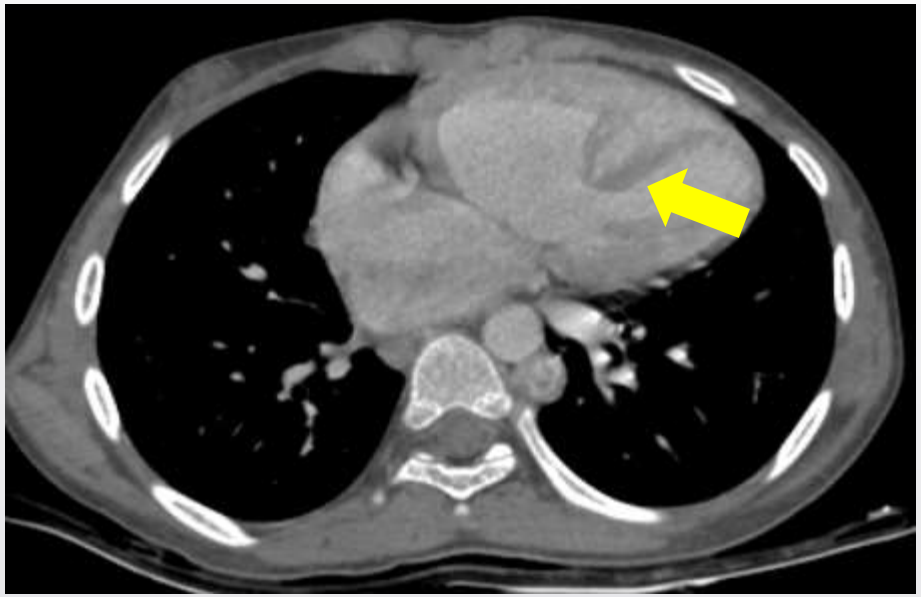
RADIOLOGICAL FINDNGS - case of left cardiac isomerism



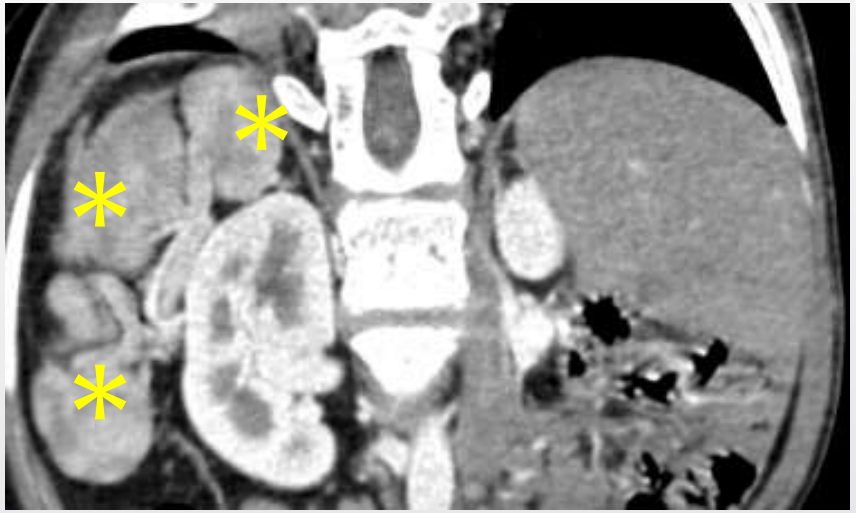
INTERRUPTED IVC WITH AZYGOUS CONTINUATION -> SVC -> RA



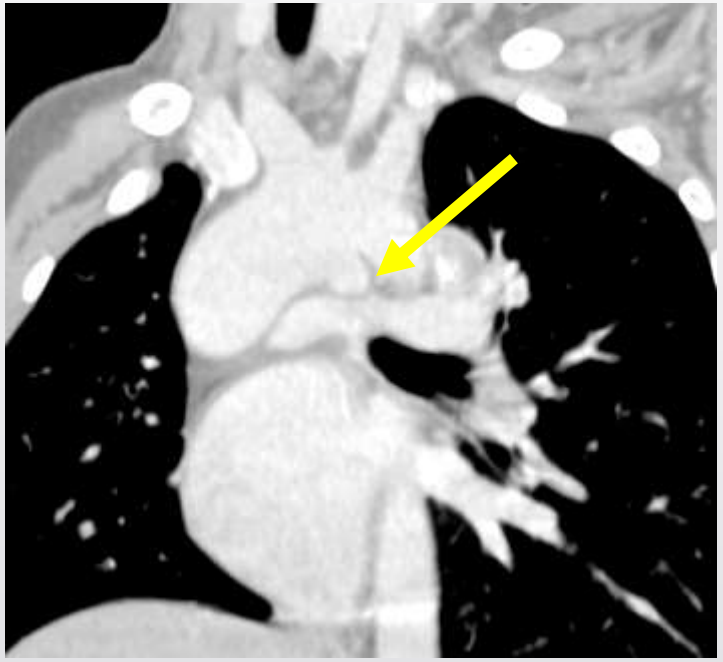
CARDIOMEGALY WITH LEFT ATRIUM & VENTRICULAR HYPOPLASIA



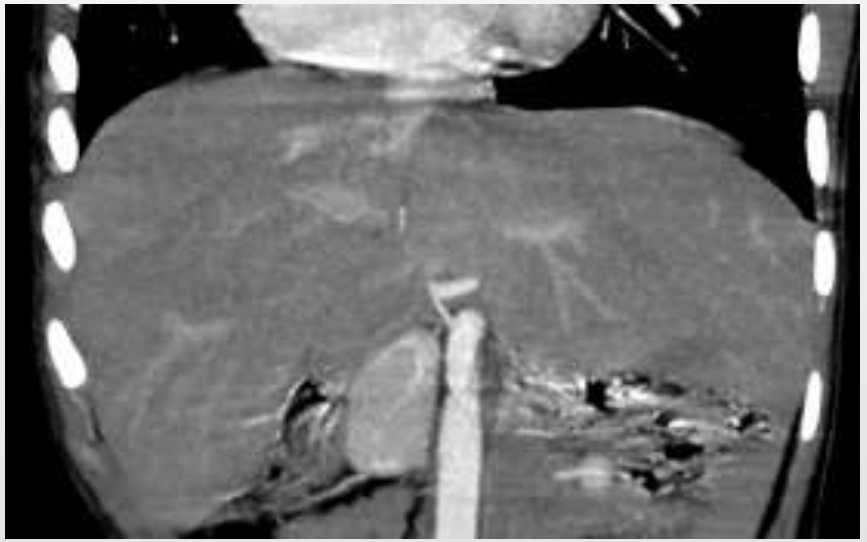
VENTRICULAR SEPTAL DEFECT



MULTIPLE LOBES OF SPLEEN – POLYSPLENIA



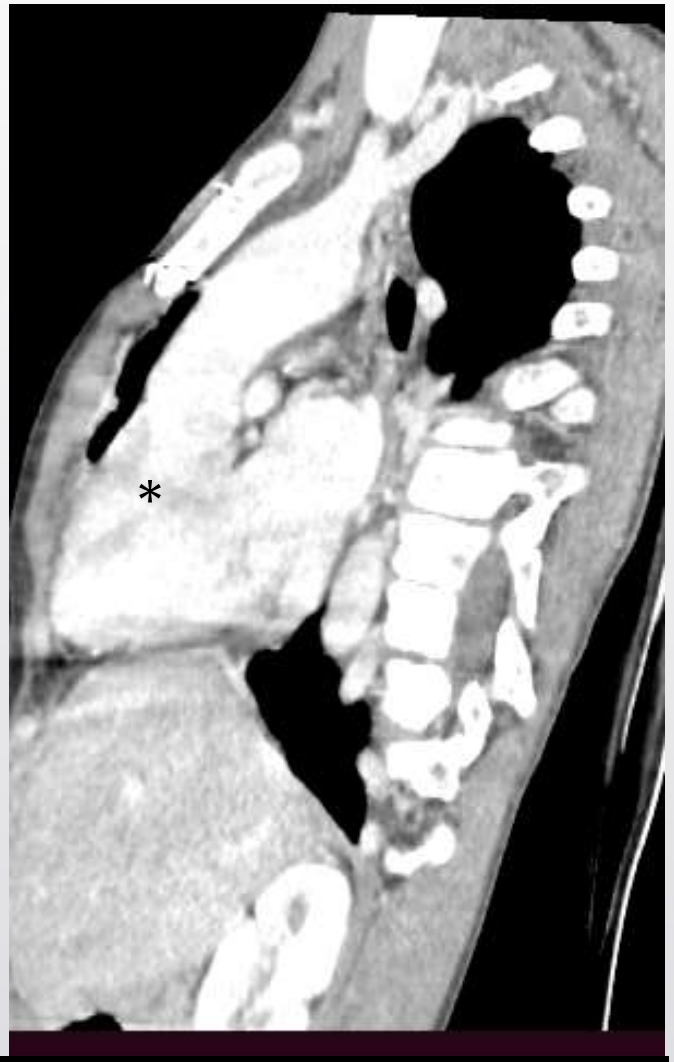
PATENT DUCTUS ARERIOSUS



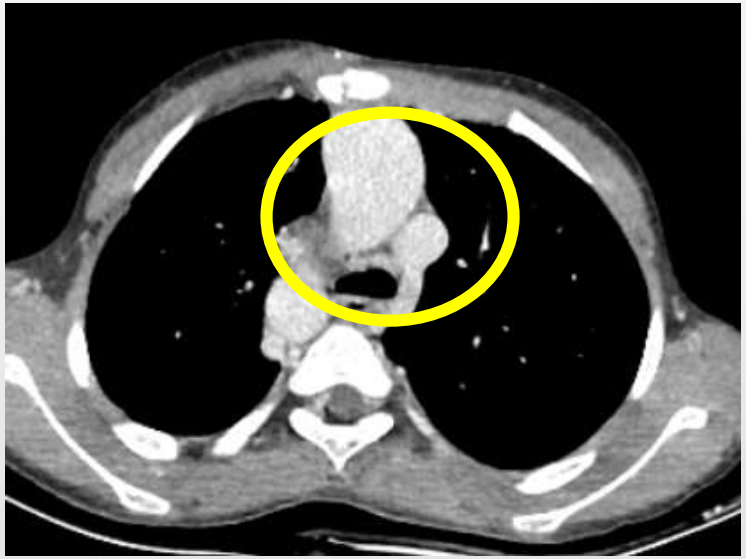
MIDLINE LIVER

- **CASE 2- 10y male, presented with recurrent history of infections**

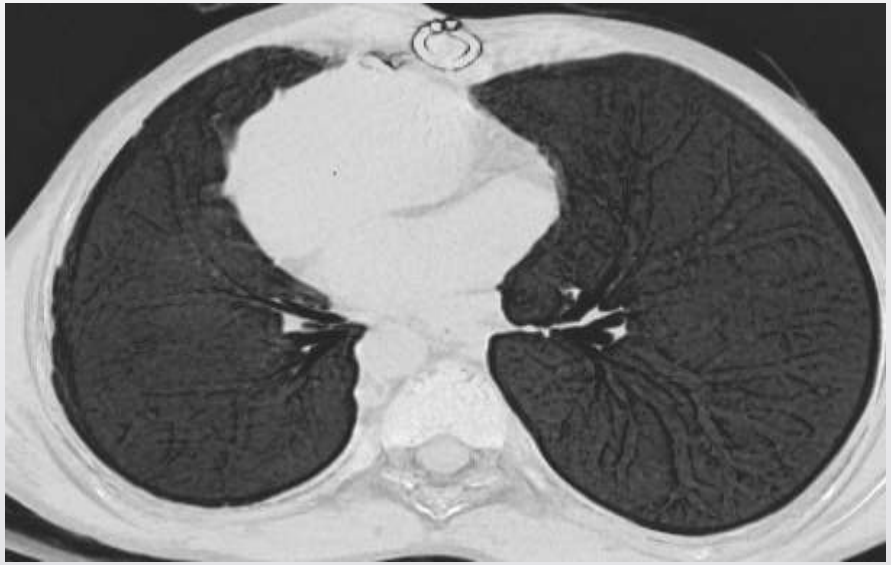
RADIOLOGIC FINDINGS : case of right isomerism



AORTA IS SEEN TO ARISE FROM MORPHOLOGICAL LEFT VENTRICLE, WHICH IS SEEN ANTERIORLY



ATRETIC PULMONARY TRUNK



CT RECON IMAGE – BILATERAL TRILOBED LUNG

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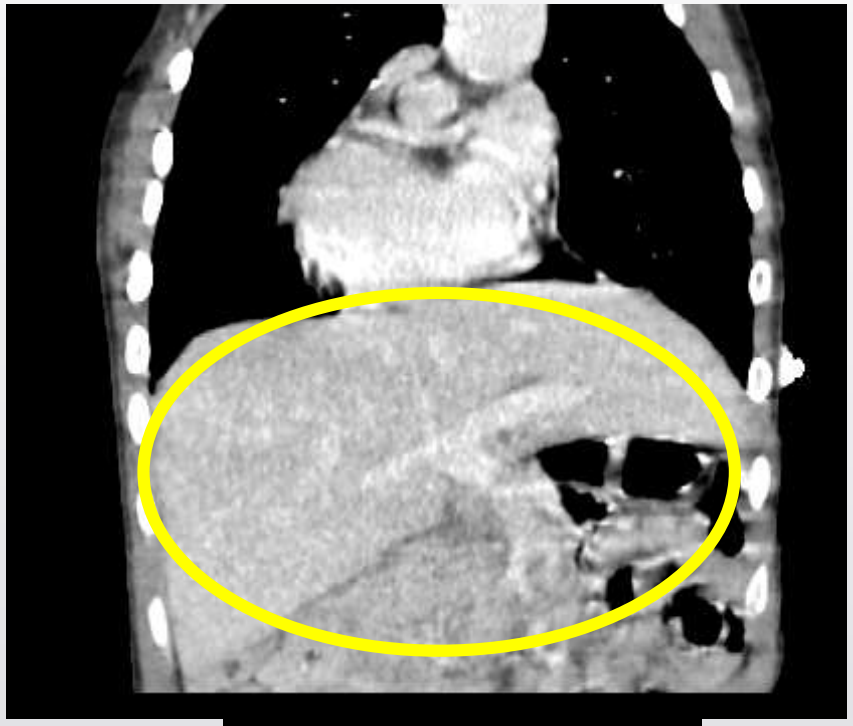
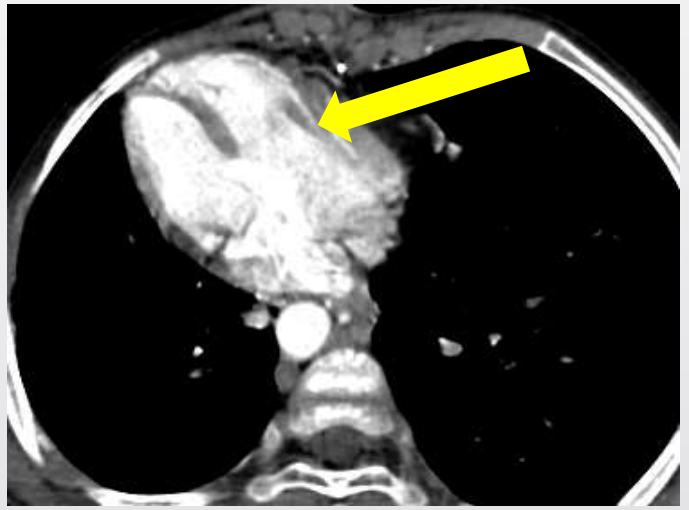


MIDLINE LIVER

ABSENT SPLEEN(ASPLENIA)



VENTRICULAR SEPTAL DEFECT

- **Heterotaxy, derived from Greek**
- ***Heteros* - other than , *taxis* – arrangement**
- ***Situs* – position**
- **Heterotaxy syndrome(HA) i.e situs ambiguous or cardiac isomerism is visceral malposition and dysmorphism associated commonly with aberrant bilateral symmetry of specific viscera, isomerism of the atrial appendage , asplenia or polysplenia.**
- **Altered development in patients of HA is considered to be related to either primary defect in lateralization of embryonic body curvature or defect in ciliary movement due to dysfunctioning calcium channels.**

- **Genetic associations i.e Pitx2, ZIC3, NODAL, CFC1, ACVR2B, LEFTY2, CITED2 and GDF1 have also been linked to heterotaxy syndrome**

- **Isomerism was broadly categorised into right and left type on the basis of splenic anatomy , i.e polysplenia and asplenia, however mere categorisation of the right and left isomerism on the basis of spleen, gives no information regarding the structural variations specially related to cardia and vessels**

COMMON FINDINGS ASSOCIATED WITH:

RIGHT ISOMERISM

- ✓ **Bilateral trilobed lungs, eparterial bronchus**
- ✓ **Bilateral isomerism of right atrial appendage- broad and round (IRAA)**
- ✓ **Liver – right or midline**
- ✓ **Splenic abnormalities - asplenia**
- ✓ **Stomach- left side**
- ✓ **intestinal malrotation (more common)**
- ✓ **Juxtaposed aorta and IVC, TAPVC and absent cornonary sinus**

LEFT ISOMERISM

- ✓ **Bilateral bilobed lungs, hypoarterial bronchus**
- ✓ **Bilateral Isomerism of left atrial appendage- thin and finger like (ILAA)**
- ✓ **Liver – left or midline**
- ✓ **Splenic abnormalities – polysplenia**
- ✓ **Stomach- right side**
- ✓ **Interruption IVC with azygous or hemiazygous continuation or IVC superior to aorta (Piggybanking appearance)**

NOTE :It is important to note that broad overlap can occur between the two type, however right cardiac isomerism is considered more severe compared to left due to reduced immunity of an individual (result of asplenia) and higher association with congenital heart disease

CONCLUSION

Congenital heart defects which are majorly associated with heterotaxy syndrome, and lead to high rates of mortality and morbidity. Widespread use and remote availability of (CT) Computed tomography machines have made it possible to diagnose heterotaxy syndrome both incidentally and in suspected individuals.

As HA, is commonly associated with diverse anomalies, delayed diagnosis can lead to worse prognosis

An early and accurate radiological diagnosis is crucial for appropriate planning of surgical and interventional procedures, as newer techniques like Fontan circulation and biventricular repair have shown encouraging results

THANK YOU