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A CLASSICAL CASE OF TURNER SYNDROME WITH POST-DUCTAL TYPE OF COARCTATION OF AORTA: SPECTRUM OF PRE-OPERATIVE IMAGING FINDINGS WITH POSTOPERATIVE STATUS

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ABSTRACT
Turner syndrome, also known as Monosomy X or 45X, is the most common of the sex chromosomal abnormalities in females. The missing chromosome is usually paternal and syndrome is not related to mothers' age. In most cases conceptions with Turner syndrome are spontaneously aborted. In this present study, the patient was an adolescent female aged 14 years (chronological age), presented to the department of endocrinology with chief complaint of delayed menarche, short stature, and decreased hearing on right side. The patient was evaluated with X-ray, USG, CT and MRI, in the preoperative state which revealed classical features of Turner syndrome, with post ductal type of Coarctation of Aorta (CoA). The clinical and imaging findings were further corroborated with karyotyping, which revealed 45XO katyotype. Patient underwent Coarctoplasty with stenting for post- Ductal type CoA with large cobalt-chromium stent- ANDRA stent.

KEYWORDS: Turner Syndrome (TS), Coarctation of Aorta (CoA), short stature, delayed menarche.

INTRODUCTION
Turner's syndrome (TS) or Ullriche Turner's syndrome is a disorder defined by an absent or structurally abnormal second X chromosome and affects around 1 in 2000 newborn females .Around one-third of the cases are diagnosed on the investigation of short stature and broad chest in mid- childhood .In most other patients, who have milder signs and symptoms of TS, the condition is diagnosed due to delayed or absent pubertal development secondary to gonadal dysgenesis either in adolescence or in adulthood.[1]

The most common etiology is complete X Monosomy (45, X). It is believed that 99% of fetuses with the classical 45, X karyotype spontaneously abort, making up 10% of all miscarriages within the first trimester.[2] The age-specific death rate in TS is around three-times higher than in the general female population.[1]

Almost 50% of patients are not diagnosed until adolescence because of primary amenorrhoea. The diagnosis of TS and thus intervention can be often delayed in children. Short stature is the trigger for the majority of the screening.[2]

CASE REPORT
The patient was an adolescent female aged 14 years (chronological age), presented with complains of short stature, generalised weakness and decreased hearing on right side. There was weakness and slowed speech since 1 month. There was history of delayed menarche, however thelarche was attained. There was no familial history of short stature/ any genetic diseases.

General Examination
On general examination, the patient had low posterior hair line, with high arched palate and posterior cleft palate. There was webbed neck, multiple nevii in the anterior chest wall, pectus exavactum deformity with shield like chest and widely spaced nipples. (figure 1-A,B,C). Overall the patient had short stature (Height-126 cm), with short arms and limbs. The weight of the patient was 28 kg {3rd centile for a 14 yr female adolescent is 32 kg.}. Further the blood pressure recorded in the upper limbs was 140/90 mmHg (High) and Pulse rate was 110/min, while in both were not recordable in the lower limbs. On cardiovascular examination sytolic murmur was noted.

The patient had history of recurrent otitis media. On examination, moderate conductive hearing loss was

recorded in the right ear, while the left ear had normal hearing. (figure 2B)

X-Ray Examination
The chest X-Ray showed sharp angular and smooth broad inferior rib notching bilaterally (figure 3). The lung fields were clear. X-Ray of the bilateral hands, AP view showed, short fourth metacarpal "positive metacarpal sign" (figure 4). There was "cubitus valgus", increased carrying angle bilaterally (figure 5).

Ultrasonography
Ultrasound of the pelvis, showed Hypoplastic uterus with streak ovaries (figure 6-A,B). On examination of the abdomen, Horse-shoe kidney was noted (figure 6C). On Renal Doppler examination, there was bilaterally increased resistance to flow in all intra renal arteries and main renal artery, that was suggestive of hemodynamic compromise . (PSV & RI: 50-60, 0.34 on right, and 30-35, 0.31 on left).

Ncct Brain: On NCCT brain, patchy areas of hypodensities were noted involving the bilateral cerebellar hemispheres, pons and the medulla. (figure 7)

Cect Chest Examination
CECT study of the chest revealed a short segment abrupt stenosis involving the aortic isthmus, 1 cm distal to the origin of the left subclavian artery. There was reduced calibre of aortic arch with dilated left subclavian artery. On 3D reconstruction, there was delineation of short segment abrupt stenosis involving the aortic isthmus, 1cm distal to the origin of the left subclavian artery. There were multiple collaterals from bilateral subclavian artery, RIMA (right internal mammary artery), LIMA(left internal mammary artery) with multiple dilated intercostal arteries. There was mildly reduced calibre of abdominal aorta.(figure 8,9)

MR Angiography
On MR angiography, it revealed abrupt stenosis 10 mm distal to the origin of the left subclavian artery with the narrowest segment measuring approx. 4 mm. (figure 10)
On 2D Echo, Left ventricular hypertrophy was noted.

Karyotyping: On karyotyping, the patient revealed 45 XO genetic makeup.(figure 11)
Based on the clinical history and findings, the constellation of imaging findings of X-ray, USG, CECT and MRI with background of a 45 XO Karyotype, the diagnosis of Turner syndrome was made.

Patient underwent Coarctoplasty with stenting for post Ductal Co A with large cobalt-chromium stent- ANDRA stent (figure 12). The pre-procedure pressure gradient at Coarcted site of 42 mmHg reduced to 5 mmHg post procedure. Patient was also started with hormone replacement therapy for activation of hypothalamo-pituitary axis.

DISCUSSION
Turner syndrome exhibit variable phenotypes, that can be split into three main categories: Monosomy X karyotype (45, X) (in 36-45%); mosaic karyotype (44-54%); and an isochromosome Xq, (5-11%).[3]

TS most commonly presents with short stature, but the phenotypical features vary and include ovarian, cardiovascular (e.g. coarctation of the aorta and bicuspid aortic valve) and renal disorders (e.g. duplicated or cleft renal pelvis; horseshoe kidney). Other features include webbed neck, broad chest with widely spaced nipples, cubitus valgus, low posterior hairline and multi-pigmented nevi. These features tend to influence the age at which the diagnosis of TS is made.[2]

CoA (Coarctation of Aorta) in Turner's syndrome are of two types: Infantile (pre-ductal) form – showing diffuse hypoplasia of the aorta from just distal to the brachiocephalic artery to the level of ductus arteriosus. The other is the Adult form (Juxta-ductal, post-ductal) –characterized by short segment abrupt stenosis of the post-ductal aorta, where the blood supply is from collateral pathway via the internal thoracic artery, posterior intercostal artery, anterior intercostal artery and then into the descending thoracic aorta. There is notching in the inferior aspect of the ribs usually the 4^{th} to 8^{th} ribs; occasionally the 3^{rd} to 9^{th} ribs.

MRI is unarguably the method of choice for visualizing the thoracic aorta. MRA using gadolinium produces clear visualization of the entire aortic arch, allowing recognition of clinically silent anomalies such as elongation of the transverse aortic arch with prominent kinking past the site of the ductus insertion (elongated trans- verse arch of the aorta [ETA]) in almost 50% of patients with TS.[1]

CT angiography plays an important role in the assessment of aortic Coarctation. The excellent spatial resolution and ease of multiplanar reformatting of CT angiography make it an excellent tool for assessing the anatomical structure of both the heart, pulmonary venous drainage, valve structure, and thoracic aorta, particularly where MRI is not available or contraindicated, although the associated radiation dose in a typically young population make this suboptimal for long- term follow-up.[1]

In TS, girls and women face a lifelong struggle with both acquired cardiovascular conditions and congenital heart disease. Bicuspid aortic valve (BAV) is common, and many have left-sided heart obstructive disease of varying severity, from hypoplastic left-sided heart syndrome to minimal aortic stenosis or coarctation of the aorta. It is becoming increasingly apparent that a variety of other cardiovascular conditions, including early-onset hypertension, ischemic heart disease, and stroke, are the major factors reducing the life span of those with Turner syndrome.[4]

A hypercoagulable state could be considered a contributing factor of thromboembolism in TS. Some case reports have described deep venous thrombosis and portal vein thrombosis in TS. Abnormalities in clotting and fibrinolytic factor levels are seen in some TS patients. These factors include fibrinogen, D-dimer, factor VIII, von Willebrand factor, and proteins C and S.[5]

Underlying arteriopathies of the cerebral arteries, including moyamoya syndrome, fibromuscular dysplasia, congenital hypoplasia, and premature atherosclerosis have been assumed to be the cause of ischemic stroke in TS.

The source of stroke in our patient remains unclear; however, this Ischemic stroke in TS could be due to embolism as well as various cerebral arteriopathies. Further investigation is needed to more fully understand the various mechanisms of ischemic stroke in TS.

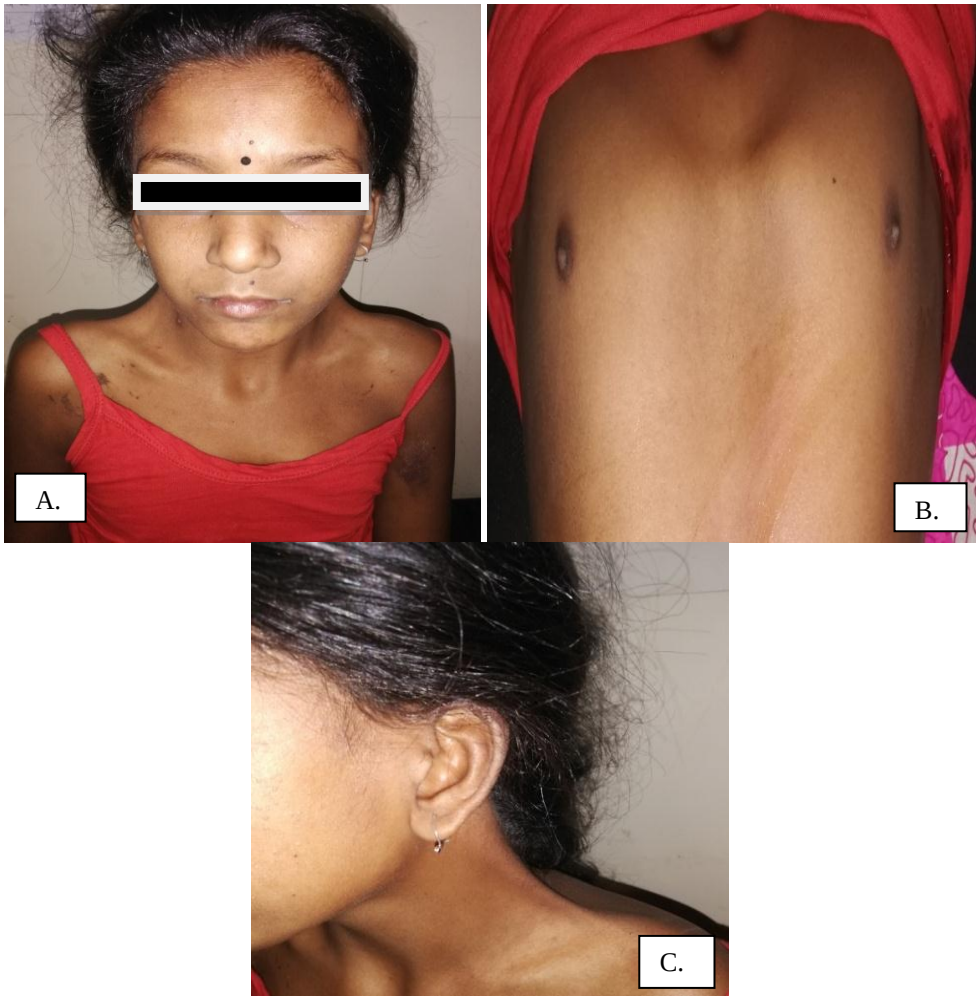
TREATMENT

Monosomy X, or Turner syndrome (TS) is the only monosomy compatible with life.(6) A multidisciplinary approach can reduce the mortality and is required for management of Turner's syndrome- surgery for cardiac anomalies, HRT (Hormone replacement therapy) for development of secondary sexual characters and fertility treatment.

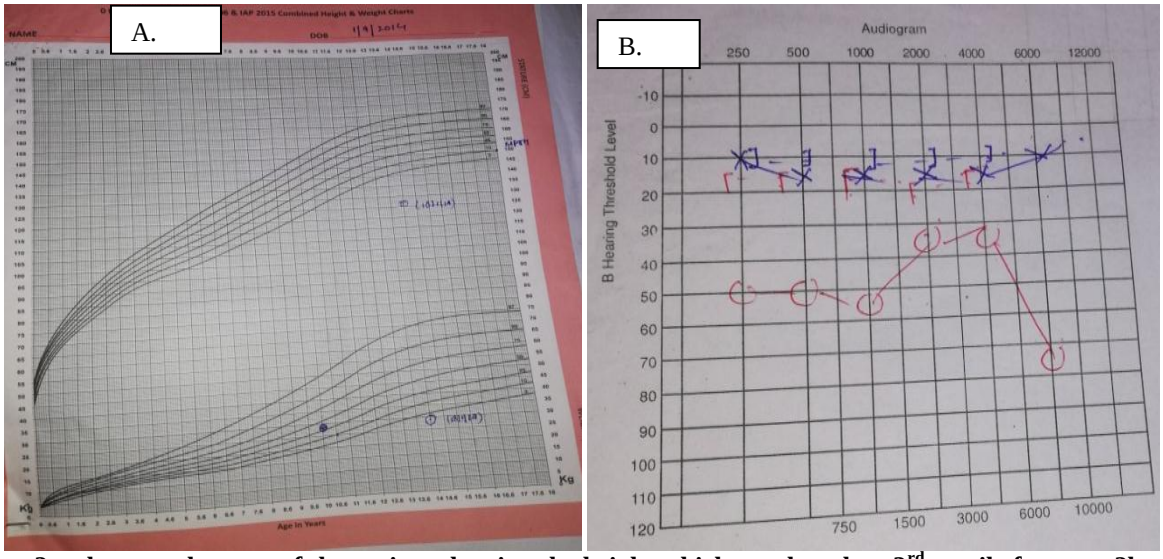
CONCLUSION

Early diagnosis of girls with Turner syndrome (TS) is essential to provide timely intervention and support.[2] With late diagnosis, puberty is already delayed and some compromises have to be made in adjusting the timing of artificially induced puberty to optimise overall outcome with respect to stature, secondary sex characteristics, and psychosocial endpoints. Further, induction of puberty regimens lack an evidence base or even clear guidelines for the timing and dose of estrogen replacement.[7]

Awareness of the common findings in Turner's, in combination with a structured approach to imaging and follow-up, will provide maximum yield in terms of early identification and management of a wide range of potentially serious cardiovascular conditions.[1]



FIGURES: 1A-Webbed neck with multiple nevi in upper chest, neck, left arm. 1B-Pectus exavactum & Shield like chest. 1C-Low posterior hair line



Figures 2a: the growth curve of the patient showing the height which was less than 3rd centile for age . 2b-on ent examination, right ear showed –moderate conductive hearing loss, while hearing in the left ear – normal.

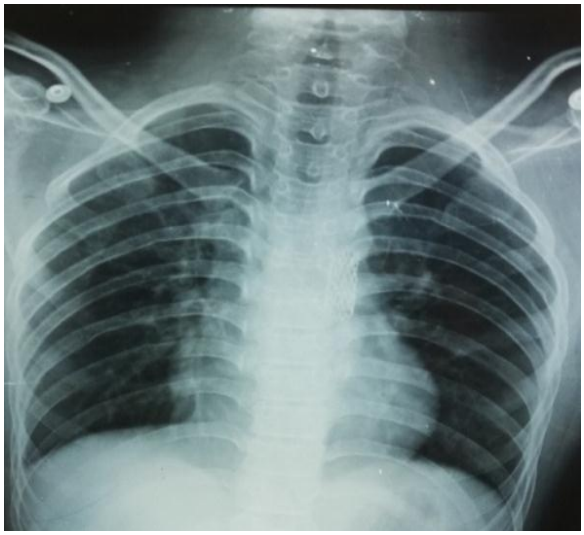


FIGURE 3: Chest X-Ray PA view of chest showing sharp angular and broad smooth inferior rib notching bilaterally.

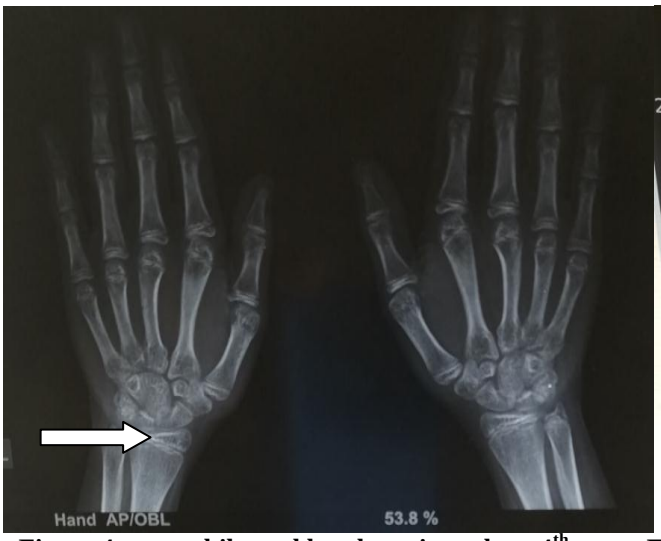


Figure 4: x- ray bilateral hand ap view: short 4th metacarpal "positive metacarpal sign".

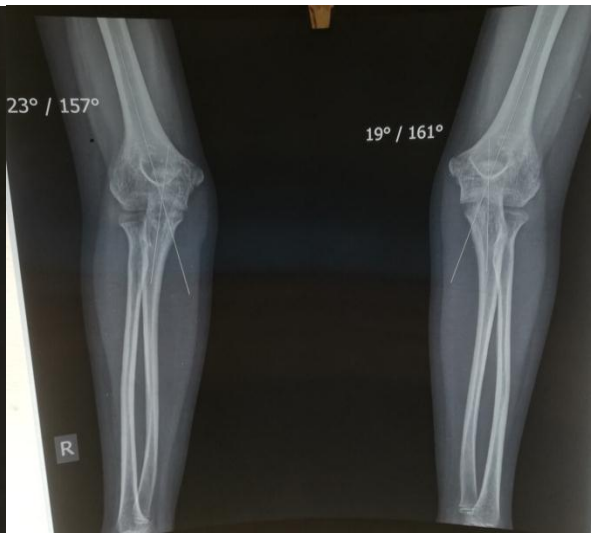


Figure 5: increased bilateral carrying angle " cubitus valgus".

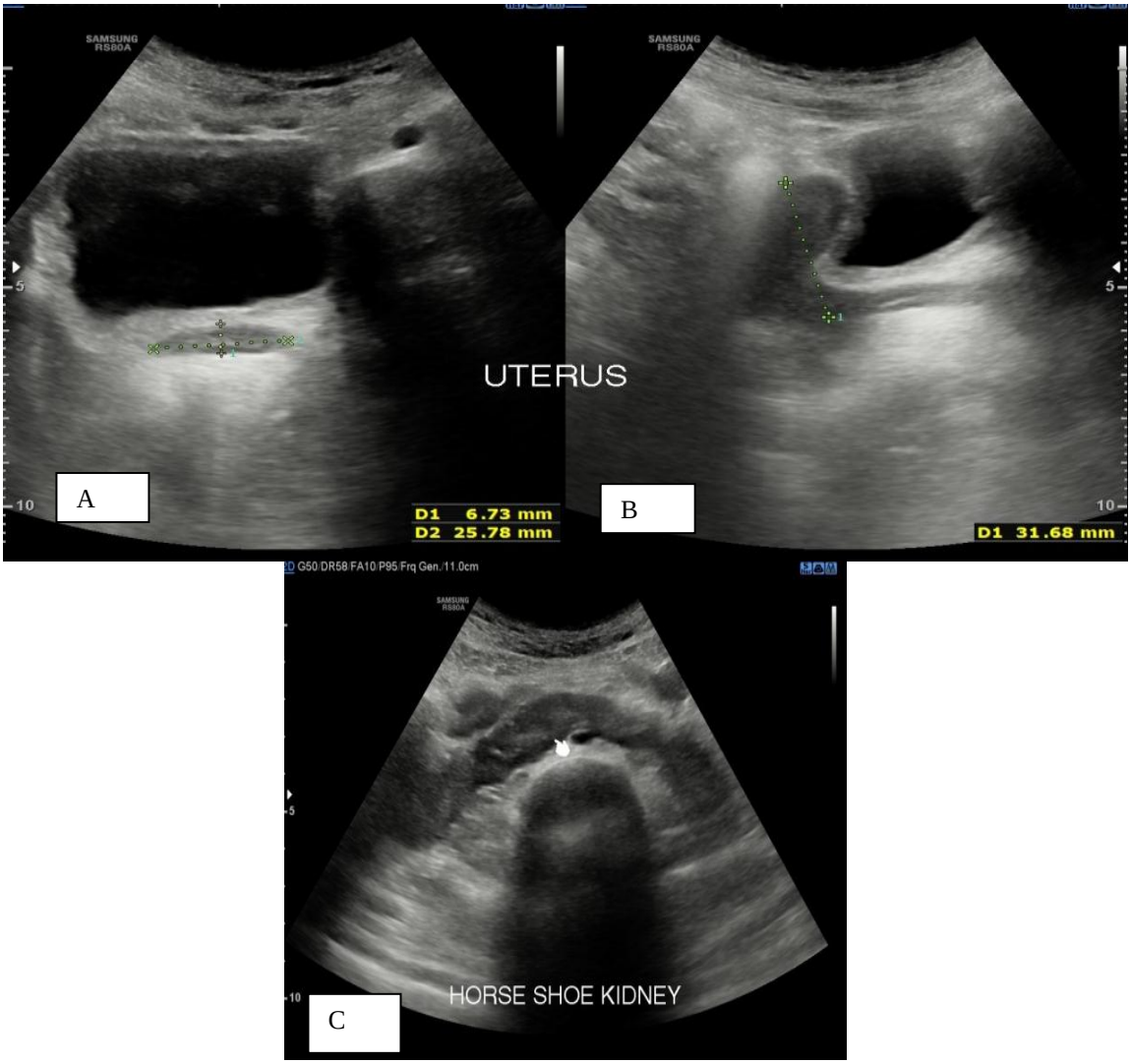


Figure 6: A, B- USG of pelvis showing Hypoplastic uterus with streak ovaries. C-Image showing " horse-shoe kidney".

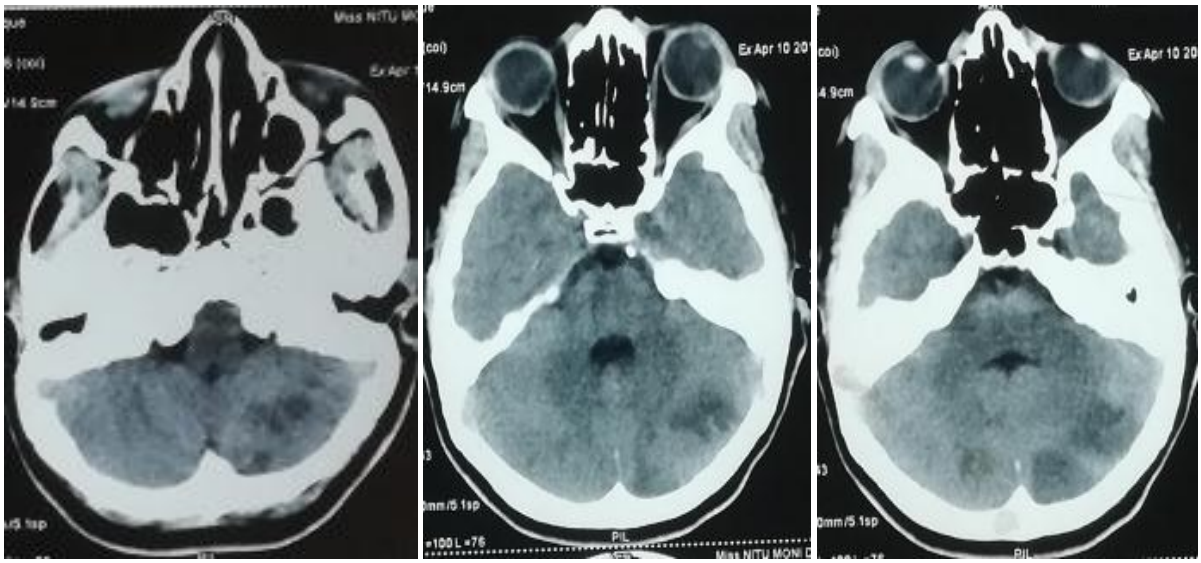


Figure 7: NCCT Axial images shows patchy hypodensities involving bilateral cerebellar hemispheres, Pons and medulla.

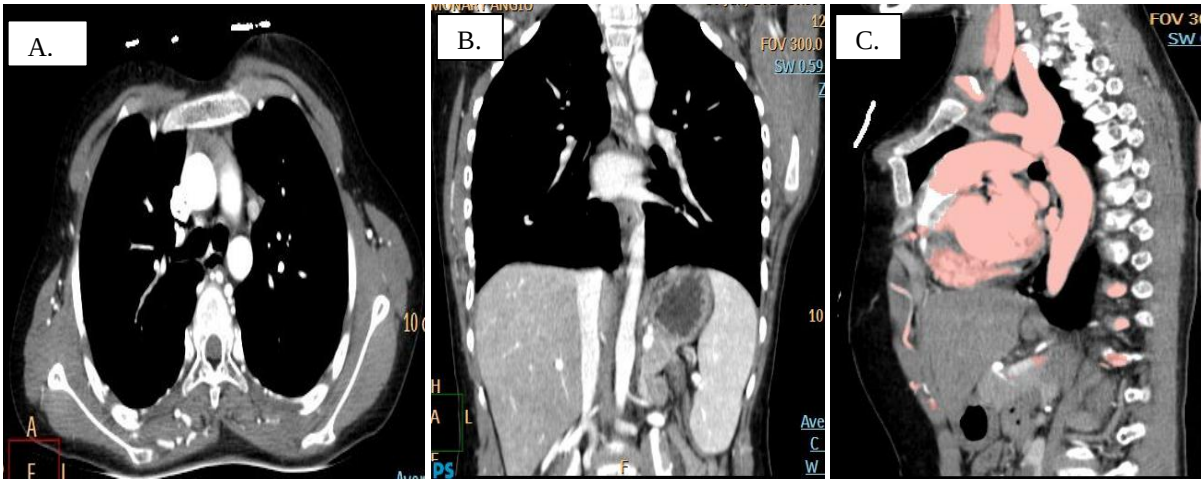


Figure 8: AXIAL, CORONAL AND THE SAGITTAL IMAGES (A,B,C)- A short segment abrupt stenosis involving the aortic isthmus, 1 cm distal to the origin of the left subclavian artery. There is reduced calibre of aortic arch with dilated left subclavian artery.

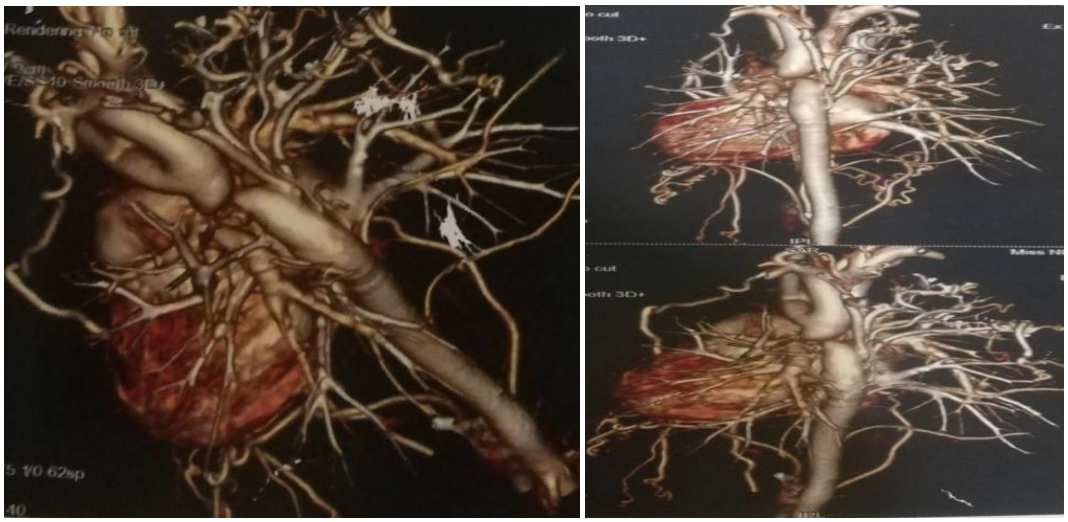


Figure 9: 3D Reconstructed images showing a short segment abrupt stenosis involving the aortic isthmus, 1 cm distal to the origin of the left subclavian artery. There are multiple collaterals noted arising from bilateral subclavian artery, RIMA, LIMA.

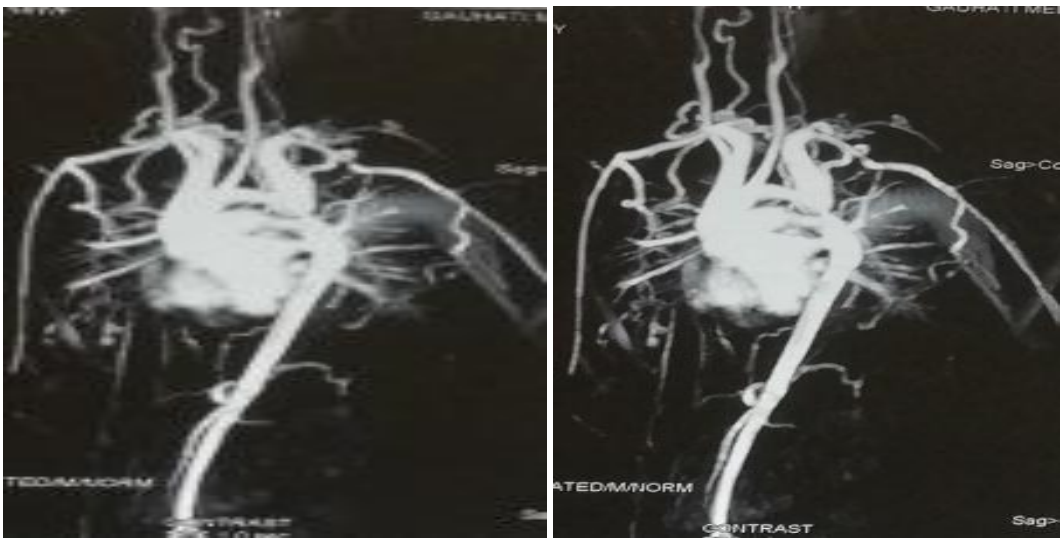


Figure 10: MR Angiography: showing abrupt stenosis distal to the origin of the left subclavian artery with the narrowest segment measuring approx. 4 mm.

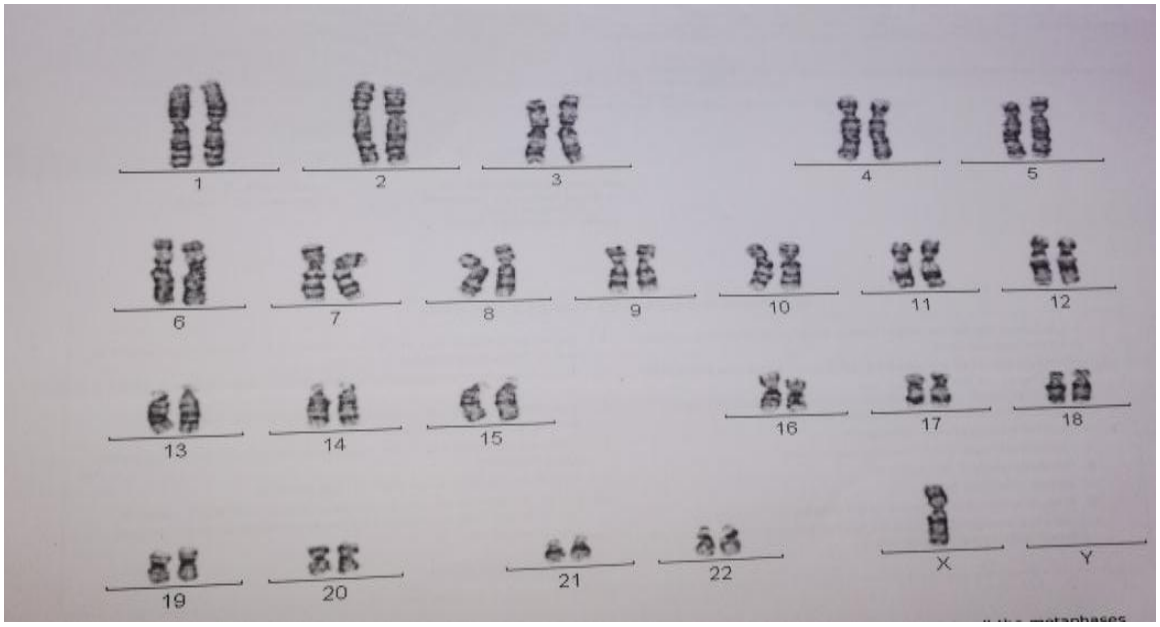


Figure 11: karyotype of the patient showing 45XO genetic makeup.

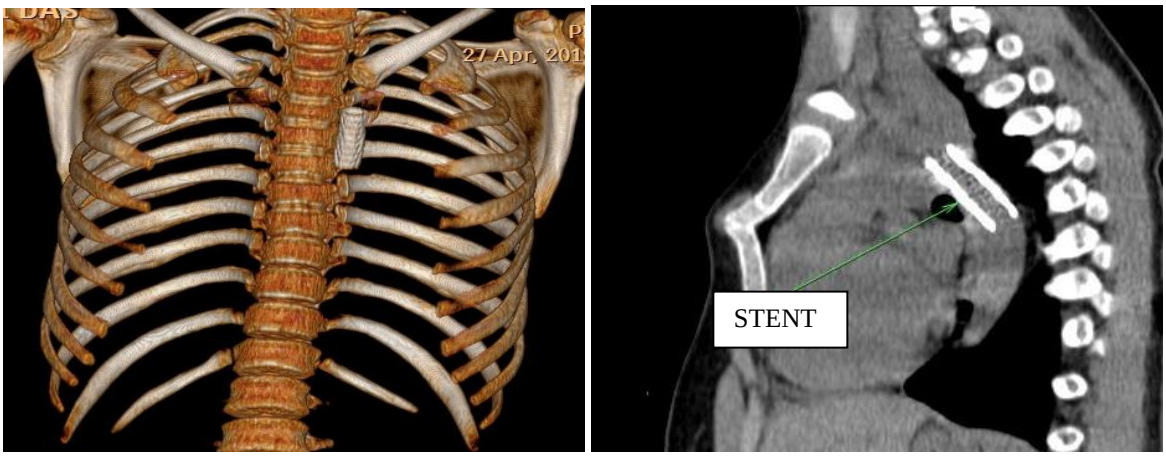


Figure 12: Post operative image showing stent in-situ. The Patient underwent Coarctoplasty with stenting for post Ductal Co A with large cobalt-chromium stent- ANDRA stent.

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