IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 21, Issue 11 Ser.5 (November. 2022), PP 46-50
www.iosrjournals.org

Arterial Tortuosity Syndrome in a 25 -Day Old neonate-A Case Report

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Abstract:
Arterial tortuosity syndrome (ATS) is rare congenital connective tissue disorder,primarily due to defect in collagen in body. It affects large and medium-sized arteries inducing tortuosity and elongation. Patients present with dysmorphic features, hyperextensibility of skin, easy bruisability,hypermobile joints, and sometimes life-threatening hemorrhages. We present a case of a 25-dayold neonate, who presented to OPD with left inguinal hernia.On routine investigations was found to have asymptomatic Arterial Tortuosity Syndrome.

Date of Submission: 06-11-2022 Date of Acceptance: 20-11-2022

I. Introduction:

Arterial tortuosity syndrome (ATS) is rare autosomal recessive connective tissue disorder, primarily due to defect in collagen in body. It is caused by a mutation in the SLC2A10 gene. It affects large and medium-sized arteries inducing tortuosity and elongation, most commonly include the aorta, arch branches and pulmonary arteries,rarely involve smaller arteries. Onset is usually in infancy or early childhood[(1).] Patients can be asymptomatic or present with wide range of manifestations. It is associated with a higher incidence of vascular complications such as aneurysm formation, vascular dissection, and pulmonary artery stenosis. Typical skeletal manifestations include dysmorphic features, hyperextensible skin, hypermobile joints, and congenital contractures[(2)]

II. Case details:

We present a 25- day old female, who presented with a complaint of swelling over the left inguinal region which increases while crying since birth. The patient also had lax hyperextensible skin.

Chest radiography showed widened mediastinum and prominent aortopulmonary window. CT aortogram was advised for further evaluation. Evaluation by CT aortogram showed gross elongation and tortuosity of ascending aorta, arch of aorta and descending thoracic aorta. Descending thoracic aorta measures approx. 1.6 cm at its origin and 0.7 cm in calibre distal to mild kinking at the level of fourth rib of posterior chest wall. No evidence of any aneurysm, dissection or thrombosis.

Tortuosity of origin of great arteries and their branches was seen leading to cluster of vessels suggestive of Cluster of vessel sign. Gross elongation and tortuosity of brachiocephalic trunk, right CCA, right subclavian artery, left CCA and left subclavian artery is noted. Gross kinking of left CCA is noted at the level of mediastinum. Both subclavian arteries show kinking in axillary region. At the areas of kinks, significant luminal narrowing is noted. Tortuous arteries seen coursing beyond the normal distribution and extending to adjacent anatomical areas suggestive of meandering vessel sign. Meandering aorta is noted upto the chest wall.

Both subclavian arteries show tortuous course with uneven calibre particularly in the axillary and mid arm region. Vascular changes are also seen in the small arteries like internal mammary arteries which show tortuous appearance. Main pulmonary artery and its branches appear elongated. Cardiomegaly with widening of subcarinal angle is seen. Tortuosity and elongation of abdominal aorta, celiac trunk, bilateral renal arteries, SMA and IMA, common iliac arteries, external and internal iliac arteries, femoral arteries and their branches is noted.

Mild depression in chest wall was seen just below sternum-Pectus excavatum

Defect measuring 1.2 cm is noted in left inguinal region with herniation of bowel.

The diagnosis of ATS was made due to typical radiological findings. Genetic or molecular testing was not done for the patient.

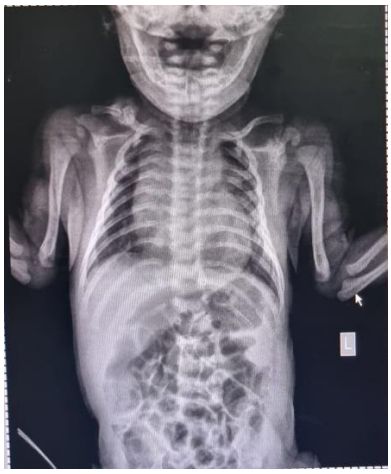

Fig.1: Mild cardiomegaly with prominent aorto-pulmonary window noted on the Chest X Ray AP view.

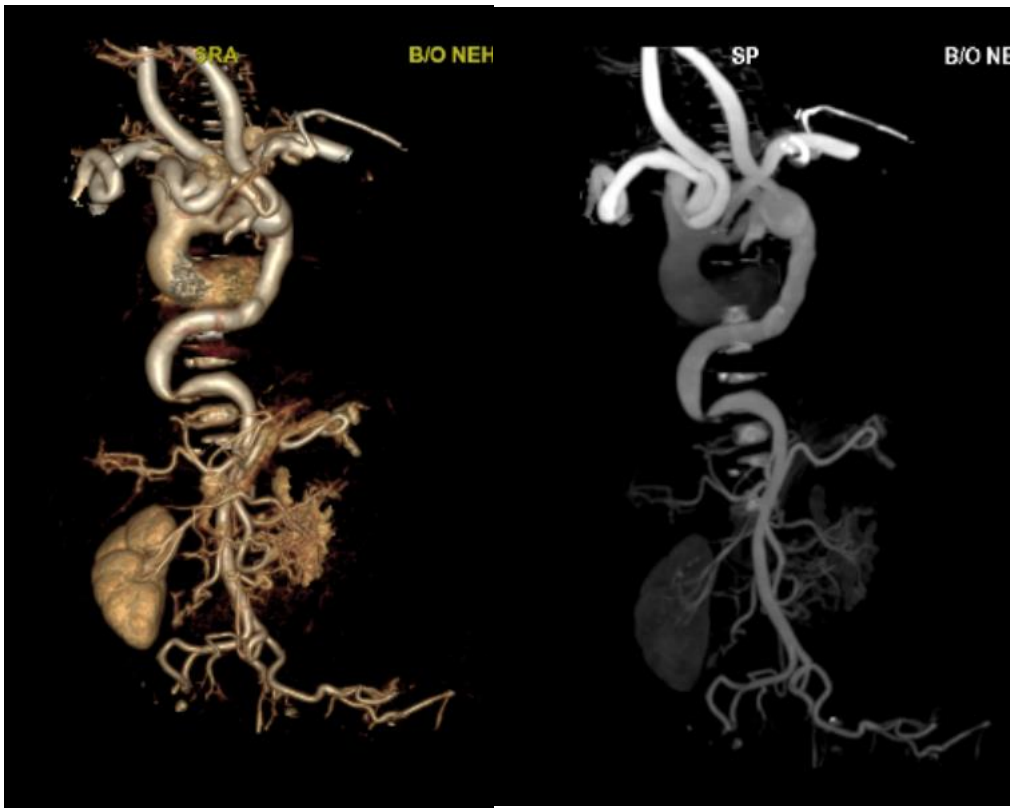

Fig2: Gross dilatation and tortuosity of thoracic and abdominal aorta**.**

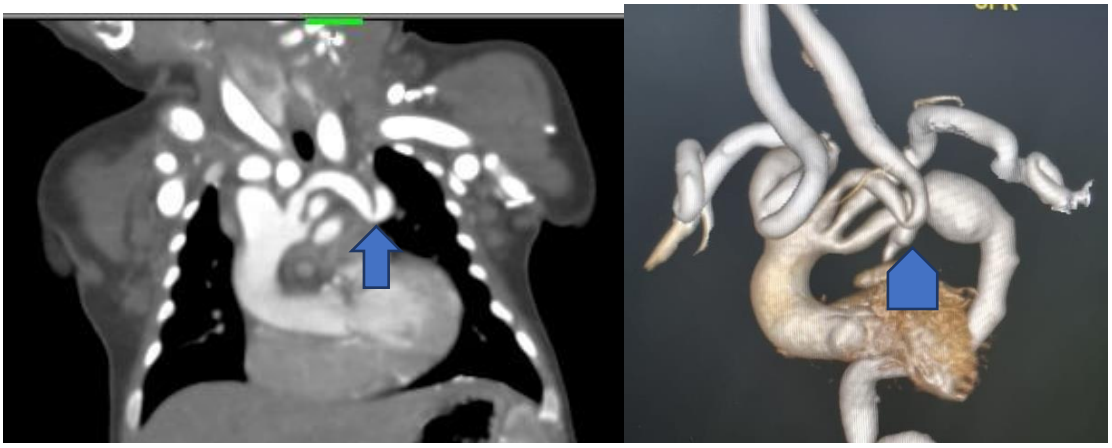

Fig3:Gross kinking of left CCA is noted at the level of mediastinum.

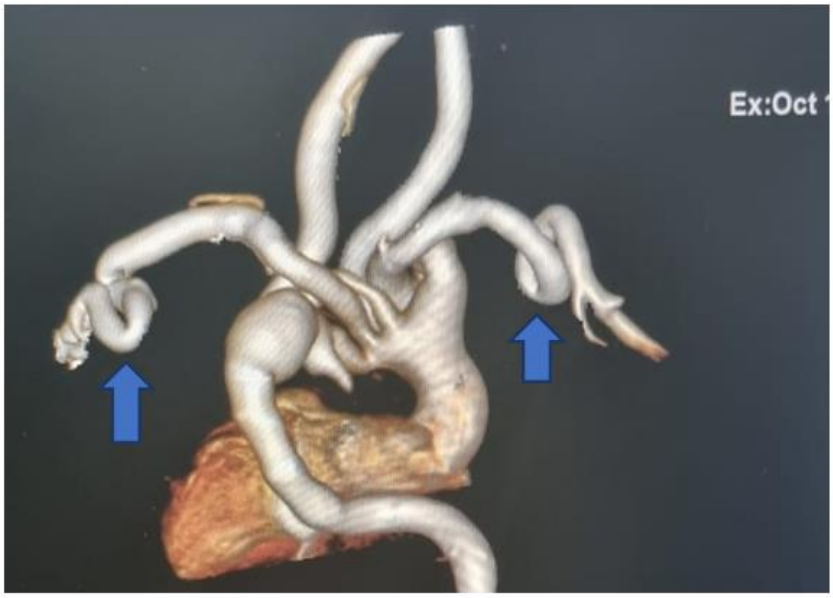

Fig 4: Both subclavian arteries show kinking in axillary region.

Fig 5:3D rendered image shows extreme tortuosity and meandering of innominate artery and left CCA-Meandering vessel sign

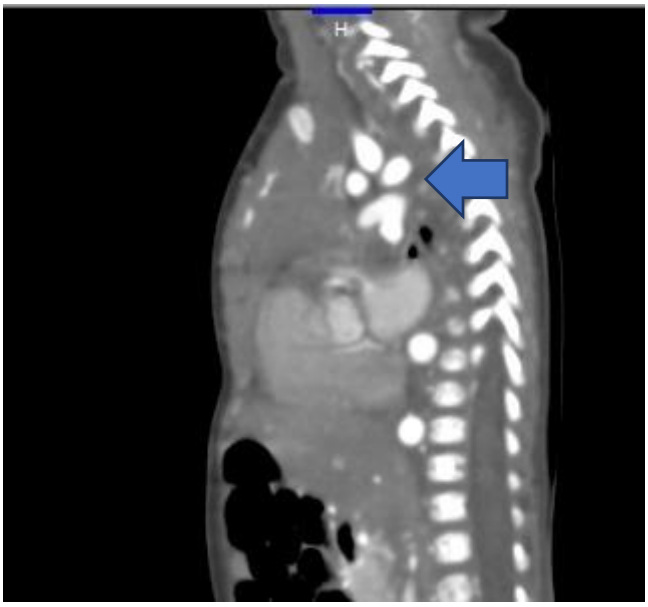

Fig 6: Tortuosity of origin of great arteries and their branches was seen leading to cluster of vessels suggestive of Cluster of vessel sign

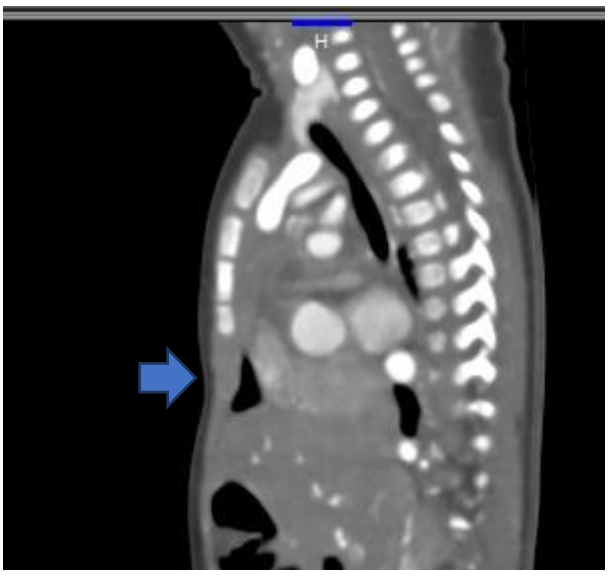

Fig 7: Mild depression in chest wall just below sternum-Pectus excavatum

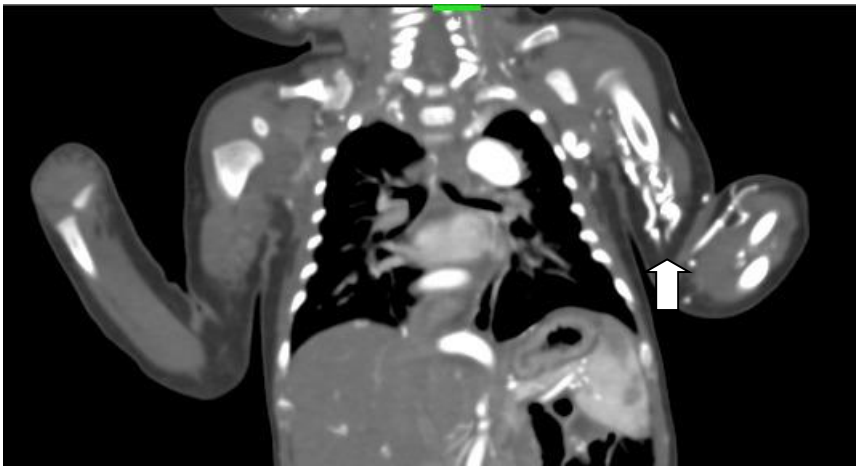

Fig 8: Multiple tortuous vessels noted in the extremities.

III. Discussion:

ATS is rare autosomal recessive disorder, characterized by disruption of elastic fibres in the medial layer of the arterial wall. The genetic defect in ATS is caused by loss-of-function mutations in the SLC2A10 gene which encodes facilitative glucose transporter GLUT 10. This is a multi-system disorder involving cardiovascular, musculoskeletal, and gastrointestinal systems[(1).]The typical cutaneous manifestation is skin laxity, which can occasionally be seen on plain radiographs. Chest radiographs demonstrate linear shadows that overlie lung fields. Skeletal manifestations include facial dysmorphism, Hyperlaxity of skin and joint, chest wall deformity as pectus excavatum and kypho-scoliosis[(2)]. Gastrointestinal manifestations include oesophageal dilatation, diaphragmatic eventration, and recurrent abdominal wall hernias[(3)]. Among the cardiovascular malformations, vascular tortuosity is the most common finding, which may be localized or generalized. Usually, it affects large vessels as the aortic arch and its branches, middle-sized vessels as coronary, cerebral, and iliac arteries, and is less likely to affect small-sized arteries. Intracranial vessels are more susceptible to multiple aneurysms. Tortuosity may attain S- or C-shaped configuration. It might have acute angulation, which is called kinking. Kinking is graded as mild (angle ≥60°) to moderate (angle between 30° and 60°) and severe (angle less than 30)[(4).] Kinking can be seen in the visceral arteries (the celiac and superior mesentery arteries).

Many syndromes have clinical similarities to ATS and should be differentiated from ATS. These include Marfan syndrome, Loeys-Dietz syndrome, aneurysms osteoarthritis syndrome, and Menkes disease. In Marfans syndrome, vascular changes often affect the aortic root in the form of aneurysm formation or dilatation.Other differentials include Loeys-Dietz syndrome and Menkes disease, autosomal recessive cutis laxa type 1[(5)]. Narrowing of pulmonary arteries is specific to ATS, as well as, diffuse involvement of the aorta, and arch branches.

IV. Conclusion:

ATS is a rare connective tissue disorder characterized by tortuosity, dilation, aneurysm, and stenosis of large and medium sized arteries. Imaging plays a vital role in diagnosis and detection of complications.

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Dr. Mannem Roopa, et. al. "Arterial Tortuosity Syndrome in a 25 -Day Old neonate-A Case Report." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 21(11), 2022, pp. 46-50.