Bilateral Renal Infarction in a Seven Months Infant Boy: Arterial Tortuosity Syndrome. A New Reported Presentation

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Abstract

The aim of this article is to report and highlight on a new fatal presentation for ATS. Arterial tortuosity syndrome (ATS) is a very rare genetic disorder, with connective tissues involvement. All over the body and it affects both sexes equally. Approximately one hundred patients have been registered in the literature. Patients with ATS have facial, musculoskeletal, genitourinary, visceral, skin and abdominal wall defects. It is associated with life-threatening complications in infancy and early childhood and the first few months of life are crucial for the high possibility of life-threatening events such as cardiac or even respiratory failure. ATS is an autosomal recessive disease due to mutation(s) in the SLC2A10 gene, and up to 23 mutations have been identified in the SLC2A10 gene by highly specific genetic investigations. The chance for a child to be genetically normal for that trait is 25%. The risk is equal for both males and females.

Keywords: Arteriomegally; Arterial tortuosity; Renal infarction

Introduction

It has been determined that SLC2A10 gene is located on the long arm (q) of chromosome 20 (20q13.1). The SLC2A10 gene encodes a protein known as facilitative glucose transporter 10 that regulates glucose and oxidized form of vitamin C, across cell membrane. Mutation(s) in SLC2A10 lead to low levels of that protein in the affected patients [1-4]. In 2015, Németh, et al. [5] conducted a study on skin fibroblasts derived from ATS patients, showed that the lack of glucose transporter 10 protein disturb the transforming growth factor beta (TGFβ) pathway and causes disorganization of different structural proteins i.e. (collagens, elastin, fibronectin) that are essential for the integrity of several connective tissues including blood vessels wall thus affecting the structural integrity of blood vessels wall specially the aorta and the pulmonary arteries.

The arterial wall is prone to elongation, tortuosity, aneurysmal dilatation, dissection and stenosis with subsequent thrombosis. The different symptoms and severity in ATS vary from one person to another depending upon the affected arterial territory. ATS usually involve large and medium sized arteries such as the aorta, the pulmonary arteries, the carotid artery, and kidney (renal) arteries [6,7]. In extremely rare cases, intracranial arteries affection was reported [8].

Case Description and Discussion

A 7 months boy was referred from pediatric cardiology department for investigational purpose and vascular consultation. General examination revealed failure to thrive after birth; he was born full term via an uncomplicated vaginal delivery to healthy parents with +ve family history of consanguinity. Birth weight was 8 pounds, head circumference was 41 cm and length was 51 cm, he had micrognathia, and bilateral inguinal hernias. All laboratory workup was in the average results.
ECG gated cardiac CT aortography:

Multiple aneurysms are identified in the root of the neck, axillary region up to 2.2 cm × 2.7 cm, with dilated, tortuosity multiple aneurysms of aortic arch branches. Aortic measurement showed aortic annulus=12 mm, sinus of valsalva=17 mm, sinotubular junction=11 mm, mid-ascending=15 mm, proximal arch=14 mm, distal arch=8.5 mm isthmus=7.5 mm, proximal descending=11.2 mm, descending aorta at diaphragmatic level=10 mm, upper abdominal aorta=9.7 mm, aorta above renal arteries=10 mm.

Brachiocephalic artery=8 mm at its origin, carotid arteries dilated up to 10 mm and ectatic left subclavian artery=9 mm. Coronary arteries revealed ectatic left main coronary up to 8.5 mm, diffuse ectasia of LAD up to 7.7 mm, and aneurysm of proximal LAD. Dilated celiac and superior mesenteric artery=8 mm and 12 mm.

Multiple focal fusiform aneurysms involving the infra-renal aorta, dilated parts reach up to 1.7 cm. Dilated tortuosity of renal arteries, diffuse, left renal artery=9 mm, diffuse non uniform dilatation of right renal artery=12 mm. Ectatic both common iliac=9 mm with common femoral reaches up 1 cm, and partial thrombosis aneurysms of internal iliac, largest on right side, 1.6 cm diameter and 3 cm in length, multifocal areas of dilatation in SFA, popliteal arteries are ectatic in both sides (12 mm × 11 mm) (Figure 2).

One week later, the boy developed severe vomiting, persistent metabolic acidosis and rising creatinine level and laboratory analysis revealed normal complete blood count, sodium (140.8) mEq/L and potassium (5.07) mEq/L, PH=7.361, PCO₂=28.9, PO₂=92.9, S. creatine=1.2 mg/dl, rising up to 1.7 mg/dl 12 hours later.

A diagnosis of arterial tortuosity syndrome is based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests and SLC2A10 gene molecular analysis, histopathology examination of affected arteries that reveal disruption of elastic fibres of arterial walls. Also diagnosis of arterial tortuosity syndrome requires a variety of specialized tests to assess the extent of the disease. Such tests include echocardiography, angiography, magnetic resonance angiography (MRA), and computed tomography (CT) scan. Molecular genetic testing confirms or excludes a diagnosis of arterial tortuosity syndrome. Molecular genetic testing can detect mutations in the SLC2A10 gene known to cause the disorder but is available only as a diagnostic service at specialized laboratories [3].

ATS [9] put the final diagnosis for ATS, on the basis of complete physical examination, thorough medical history evaluation, Assessment of signs and symptoms, Laboratory tests, Imaging studies, Biopsy studies, if necessary.
The diagnosis of our reported case of arterial tortuosity syndrome was based on characteristic symptoms, signs, the detailed patient history, +ve family history of consanguinity, clinical evaluation and angiography finding that revealed dilatation, tortuosity of both renal arteries, large arteries affection. (Angiography report) together with bilateral wedge shaped hypodense areas in both kidneys (CT scan report). Colour duplex and renal ultrasound revealed thrombosis of both renal arteries and wide areas of infarction (Figure 3). Molecular genetic testing was not feasible in our patient due to lack of such specialized tests in our genetic unit.

Finally, the treatment of arterial tortuosity syndrome is directed toward the specific symptoms that are encountered in each patient. Treatment needs coordinated efforts of a team of different specialties. Psychosocial support and assurance for the parents, our reported case is under strict follow up, and supportive measures that included anticoagulation (low molecular weight heparin). LMWH was administered with twice-weekly monitoring. The median duration of therapy with LMWH was 14 days and was given in 1.0 mg/kg. Endovascular thrombolytic therapy as an urgent treatment was not feasible due to anatomical and laboratory considerations for this particular case, and unfortunately follow up creatinine level reached 2.2 mg/dl and we started hemodialysis through duplex guided right internal jugular catheter and our case is still under hemodialysis and the last creatinine level was 3.12 mg/dl.

References

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