Unravelling the Mystery of Arterial Tortuosity Syndrome: A Case Report of an Unexpected Rare Diagnosis in a Well-Baby Clinic at a Primary Care Center in Qatar

Maryam Mohamed AlDosari

Manager of Health Centre & Specialist Family Medicine Operations - HC Al Wakra (South) Qatar

Correspondence:
Dr. Maryam Mohamed AlDosari
Manager of Health Centre & Specialist Family Medicine
Operations - HC Al Wakra (South)
P.O. Box 26555 | Doha | Qatar
Qatar
Tel: 402-75701
Email: mmaldosari@phcc.gov.qa

Received: Febuary 2024. Accepted: March 2024; Published: April 1, 2024.
Citation: Walaa Abdulmohsin. Unravelling the Mystery of Arterial Tortuosity Syndrome: A Case Report of an Unexpected Rare Diagnosis in a Well-Baby Clinic at a Primary Care Center in Qatar. World Family Medicine. April 2024; 22(4): 43-45. DOI: 10.5742/MEWFM.2024.95257637

Abstract

A case of Arterial Tortuosity Syndrome (ATS) is a rare autosomal recessive genetic disorder characterized by significant alterations in the structure and function of the arterial system. Mutations in the SLC2A10 gene, responsible for encoding the transporter protein GLUT10, lead to the development of ATS.

Keywords:
Arterial Tortuosity Syndrome, case report, well baby clinic, Qatar

Background

Arterial Tortuosity Syndrome (ATS) is a rare autosomal recessive genetic disorder characterized by significant alterations in the structure and function of the arterial system. Mutations in the SLC2A10 gene, responsible for encoding the transporter protein GLUT10, lead to the development of ATS. The syndrome is characterized by the elongation, twisting, and increased tortuosity of arteries, predisposing patients to a plethora of vascular complications, including aneurysms, stenosis, and tortuosity [1]. Adding to that, ATS affects connective tissues across various systems, leading to special facial features that can be recognizable at birth or later during early childhood [2]. Being a rare and complex disorder signifies the importance of raising awareness among healthcare professionals, particularly in settings where genetic disorders might not be the first consideration such as well baby clinics where the parents bring their children just for routine well baby follow up and vaccines.

Rationale

The diagnosis of ATS in a primary care setting is exceptionally rare, given the complex presentation and the need for specialized genetic testing for confirmation. This case report aims to highlight the role primary care physicians can play in the early identification and referral of cases suspected of having rare genetic disorders in general and AST in particular. Sharing experience and insights from this unique case can contribute to the broader medical literature by underlining the necessity for vigilance and a comprehensive approach in the evaluation of infants and young children presenting with nonspecific symptoms and dysmorphic features.

Case presentation

In June 2022, a 2-month-old baby boy was brought by his mother to a well-baby clinic for a routine check-up and vaccination. The boy weighed 5.4 kg and was 61 cm tall. The mother expressed concern over the baby's excessive crying. The maternal history included gestational diabetes mellitus (managed without insulin) and recurrent urinary tract infections. The prenatal history was insignificant; all fetal ultrasounds were normal. The baby, born at a gestational age of 38 weeks to a 31-year-old G5P3 mother via elective caesarean section, did not require resuscitation, with Apgar scores of 9 and 10 at 1 and 5 minutes, respectively. The neonatal period was marked by jaundice (2).

The family's social history noted non-smoking, first-degree relative parents with secondary school-level education. The family consisted of the father (32 years old), the mother (31 years old), two girls (aged 10 and 5), and two boys (aged 8 and the 4-year-old patient). The patient's growth and milestones were normal according to the WHO pediatric growth charts. However, examination revealed dysmorphic features such as low-set ears, a high arched palate, and wide epicanthic folds, though these observations were not observed by the mother. The chest examination showed abnormal shape and pectus excavatum but normal heart sounds without murmurs. The abdomen appeared distended during crying, and the neurological examination revealed hypotonia but no limb deformities. The genitourinary examination was normal. A follow-up plan was agreed on with the mother, with a referral to pediatric emergency services for further assessment and possible referral to genetics.

Investigations

No special investigations were done at primary care as possible genetic tests can only be done through the pediatrics department. The referral aimed to exclude serious conditions and initiate further investigations for an accurate diagnosis.

Differential diagnosis

Initial differential diagnoses included diaphragmatic hernia, various connective tissue disorders, Klinefelter syndrome, and Arterial Tortuosity Syndrome.

Treatment

The treatment plan included vitamin D3 supplementation, antipyretics as needed, and comprehensive health advice to the parents which were provided based on age and routine well baby visit, but nothing was prescribed based on the accidental findings for which the baby was referred to pediatrics.

Outcome and Follow-up

The follow-up protocol included well-baby primary health care center visits at 4 months old as per the protocol and referrals to secondary care for genetic studies that reveal gene mutation (SLC2A10 Homozygous state) and pediatric services, like Neurology to follow up the Axial hypotonia , pulmonology to follow up the moderate to severe pectus excavatum and provide the prophylactic medicines, urology to follow up the left proximal hydroureter and pelviectasis, pediatric surgery for the bilateral inguinal hernia and cardiology as risk assessment and follow up. The patient also received rehabilitation services at a child development center and ongoing well baby visits are planned.

Discussion

This case report is an example of the difficulties and possibilities associated with diagnosing uncommon genetic disorders, like ATS, in a primary care setting when the presentation was made for an unrelated reason and the family were not concerned about the disorder's symptoms. Although it is rare, the early identification of ATS highlights the significance of considering a wide variety of differential

diagnoses when dealing with such clinical presentations. In this instance, the multidisciplinary approach combining pediatricians, cardiologists, and geneticists is essential, supporting the need for integrated care for such uncommon genetic disorders. This case does also support the value of genetic testing and the necessity for medical professionals to be competent identifying these features early.

Lastly, this case report highlights the need for primary care practitioners to be aware of and consider rare genetic disorders in their differential diagnosis, facilitating timely referral and appropriate management.

Ethical considerations

PHCC IRB approval was obtained. This case report addresses the principle of beneficence, as outlined in the Belmont Report, by prioritizing the well-being of the patient through early diagnosis and intervention, thus aiming to prevent possible complications. Privacy and autonomy were maintained by ensuring confidentiality and consenting parents.

References

1- Coucke PJ, Willaert A, Wessels MW, et al. Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nat Genet. 2006;38(4):452-457.
2- Callewaert BL, Willaert A, Kerstjens-Frederikse WS, et al. Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. Hum Mutat. 2008;29(1):150-158.