CARE Checklist (2013) Statement Table

Title of Case Report: Multidisciplinary Management of ATS in a Pediatric Patient: a case report
Target Journal Submission Document: CARE Checklist Compliance Statement

Item No.	CARE Checklist Topic	Extracted Text / Statement from Manuscript	Section / Page Reference
1	**Title**	"Multidisciplinary Management of ATS in a Pediatric Patient: a case report"	Title / Page 1
2	**Key Words**	"Arterial Tortuosity Syndrome; pediatric vascular disease; SLC2A10 mutation; vascular tortuosity; respiratory distress; congenital vascular anomalies; failure to thrive; GLUT10 deficiency; connective tissue disorder."	Keywords / Page 1
3a	**Abstract: Introduction**	"This case demonstrates the phenotypic heterogeneity and diagnostic complexity of ATS. Although cardiovascular abnormalities are considered hallmark manifestations, recurrent respiratory distress and failure to thrive were prominent features in our patient, highlighting that substantial pulmonary morbidity may be an important component of the early clinical presentation."	Abstract / Page 1
3b	**Abstract: Symptoms / Clinical Findings**	"We report the case of a 14-month-old Jordanian male who	Abstract / Page 1

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		presented to the emergency department with recurrent respiratory distress, failure to thrive, and cyanotic episodes... Physical examination revealed multiple connective tissue and dysmorphic features, including hypotonia, hernia, hyperextensible skin, talipes equinovarus, and blue sclera."	
3c	**Abstract: Diagnoses & Interventions**	"Genetic testing identified a homozygous frameshift variant in SLC2A10 (c.56dup; p.(Leu20Profs*6)), establishing the diagnosis of ATS. Imaging and echocardiographic studies additionally demonstrated cardiopulmonary involvement consistent with the syndrome, necessitating multidisciplinary evaluation and management involving cardiology, pulmonology, and cardiothoracic surgery."	Abstract / Page 1
3d	**Abstract: Conclusion**	"Early recognition is essential because delayed diagnosis may result in clinical deterioration and missed opportunities for appropriate	Abstract / Page 1

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		cardiovascular and pulmonary surveillance. This case emphasizes the importance of considering ATS in infants presenting with connective tissue features accompanied by recurrent respiratory manifestations and supports early genetic testing, multidisciplinary care, timely intervention, and appropriate surveillance to improve clinical outcomes."	
4	**Introduction / Background**	"Arterial Tortuosity Syndrome (ATS) is a rare autosomal recessive connective tissue disorder caused by biallelic pathogenic variants in the SLC2A10 gene, resulting in dysfunction of the GLUT10 transporter. It is characterized by elongation and tortuosity of large- and medium-sized arteries, with possible aneurysm formation and stenosis... Children during infancy usually present with dyspnea and cyanosis due to involvement of the pulmonary arteries. Several patients died at a young age, mainly because of cardiopulmonary	Background / Page 1

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		complications."	
5a	**Patient Information: Demographics**	"A case of a 14-month-old Jordanian male admitted to the pediatric intensive care unit (ICU)..."	Case Presentation / Page 1
5b	**Patient Information: Main Symptoms / Concerns**	"...suffering from bilateral pneumonia, poor feeding, gastroenteritis, and failure to thrive. His medical history included difficulties in feeding for a long time, inadequate weight gain since birth, and multiple episodes of cyanosis during feeding and crying."	Case Presentation / Page 1
5c	**Patient Information: Medical / Family History**	"His medical history included difficulties in feeding for a long time, inadequate weight gain since birth... Immunizations are up to date. His family history does not include congenital diseases or consanguinity."	Case Presentation / Page 1
6	**Clinical Findings**	"During physical examination, the patient appeared ill, hypoactive, febrile, and mildly dehydrated with oxygen saturation at 88% on room air. He was underweight and malnourished... tachypnea, flaring nostrils, a congested throat, intercostal retractions, a barrel-shaped chest, diminished breath	Case Presentation / Page 1–2

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		sounds, and cyanosis of the face (lips) and fingertips... abnormal cheek morphology, blue sclerae, excessive wrinkling of palmar skin, hernia, hypotonia, premature skin wrinkling, and talipes equinovarus."	
7	**Timeline**	- **Birth – 14 Months:** Inadequate weight gain, long-term feeding difficulty, cyanotic episodes during crying and feeding. - **Presentation (14 Months):** Acute emergency admission to PICU with bilateral pneumonia, gastroenteritis, hypoxia (88% room air), and severe respiratory distress. - **In-hospital Stay:** Physical examination identifying CTD features, genetic confirmation of *SLC2A10* variant, diagnostic imaging (CXR, ECHO, US), and multidisciplinary consultations. - **Post-Stabilization:** Discharge planning with outpatient pediatric cardiology and cardiothoracic surgery follow-up.	Case Presentation / Page 1–2
8a	**Diagnostic Assessment: Methods & Results**	"The patient's genetic test result showed a homozygous, presumed	Case Presentation / Page 1–2

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		loss-of-function variant in the SLC2A10 gene (c.56dup p.(Leu20Profs*6)). Chest X-ray revealed lung hyperinflation, increased interstitial markings (pulmonary edema), enlarged cardiac silhouette, and mediastinal shift. Echocardiograph found blood flow, valves, aortic and pulmonic vessels to be normal with the exception of anomalous venous drainage, a small patent foramen ovale (PFO), and a small ventricular septal defect (VSD)."	
8b	**Diagnostic Assessment: Challenges**	"A molecular genetic test should be performed to distinguish ATS from other connective tissue diseases (CTDs), as it shares some dysmorphic features with them, such as Marfan syndrome (MFS), Loeys-Dietz syndrome (LDS), and Ehlers-Danlos syndrome (EDS)."	Background & Discussion / Page 1–2
9	**Therapeutic Intervention**	"The pulmonary team management plan was to stabilize the patient on continuous-flow low-oxygen therapy to maintain his oxygen saturation above 92%. His vital signs and	Case Presentation / Page 2

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		respiratory status were monitored regularly... Needs were referred to cardiology for management: routine echocardiography and oxygen therapy. Potential surgical interventions were evaluated by the cardiothoracic surgery team..."	
10	**Follow-up and Outcomes**	"After patient stabilization, discharge planning began with outpatient follow-up with pediatric cardiology and surgical teams to discuss potential surgical interventions once the patient could tolerate them."	Case Presentation / Page 2
11a	**Discussion: Strengths & Limitations**	**Strengths:** "Our unique case report talks about a young child who presented to the ER with pneumonia and was diagnosed with ATS syndrome after doing genetic test... Laboratory genetic testing for this patient revealed a new SLC2A10 gene mutation called c.56dup p.(Leu20Profs*6)... In contrast to the common presentation of the ATS, this case highlights its heterogeneity and shows that significant respiratory morbidity	Case Presentation & Discussion / Page 1–3

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		may occur even in the absence of severe structural cardiovascular abnormalities early in life." **Limitations:** Primary limitations include short initial follow-up duration prior to surgical intervention tolerance.	
11b	**Discussion: Literature Comparison**	"ATS is a rare autosomal recessive genetic condition... characterized by elongation, aneurysm, stenosis, and tortuosity of the major arteries... Loss of SLC2A10 function activates the TGF-β signaling pathway... Other characteristics of ATS are similar to those of CTDs, such as MFS, LDS, and EDS... Major ATS complications that led to a 40% mortality rate in patients under 5 years include cardiovascular system, respiratory insufficiency, or ventricular hypertrophy..."	Discussion / Page 2–3
12	**Patient Perspective**	"The nature of the ATS and the anticipated long-term prognosis were also discussed with the family."	Case Presentation / Page 2
13	**Informed Consent**	"Consent to participate: Written informed consent to participate in the study was obtained	Statements and Declarations / Page 3

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		from the child’s parent. Consent for publication: Written informed consent was obtained from the patient's parent for publication of this case report and any accompanying images."	