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Case Report

Takayasu Arteritis in a Patient with Down Syndrome: A Case Report

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Abstract: Takayasu arteritis is a chronic vasculitis of large- and medium-sized vessels that primarily involves the aorta and its proximal branches. It is an uncommon disease that occurs predominantly in young women. Histopathologically, the affected vessels are characterized by mononuclear infiltrate and granulomatous inflammation of the tunica media, leading to arterial wall thickening, with stenosis, occlusion, and aneurysmal dilation. The present article aims to report on a case of Takayasu arteritis in a patient with Down syndrome.

Keywords: Takayasu Arteritis; Down Syndrome; Carotid Stenosis; Case Report.

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1. Introduction

Takayasu arteritis (TA), also known as "young woman's arteritis" or "pulseless disease" [1], is a chronic inflammatory disease classified as a granulomatous vasculitis of large vessels, involving the aorta and its main branches, including the renal, carotid, and subclavian arteries [2]. It is a disease with high morbidity [3], which may cause occlusion, stenosis, dilation, and the formation of arterial aneurysms [4]. This condition is rare and mainly affects young adults, especially women, with differences in its geographic distribution and a higher prevalence in Asian regions, suggesting a possible genetic correlation [1,3].

In this pathology, fibrinoid necrosis of the vessel wall may occur, and its pathophysiological mechanism has not yet been fully elucidated. The clinical presentation depends on the affected region, and disease progression occurs in three phases. The first phase is characterized by nonspecific symptoms such as fever, arthralgia, and anorexia; this is the inflammatory phase, in which inflammatory cells begin to migrate to the arterial media layer through the vasa vasorum. In the second phase, there is amplification of the inflammatory process, with pain along the course of the affected vessel and progression of inflammatory cells to the intima, leading to destruction of collagen fibers, resulting in the onset of stenotic areas, arterial dilation, and even occlusion. The final stage of disease progression, the third phase, is characterized by fibrosis of the arterial wall, culminating in stenosis, occlusion, and arterial dilation, thus leading to symptoms secondary to ischemia, such as claudication of the affected arterial territory and pulse asymmetry, among others [4–7].

The diagnosis of this condition is challenging; however, the American College of Rheumatology proposed criteria to facilitate the diagnostic process. These include clinical and imaging criteria [8]. Doppler ultrasonography is a valuable tool for identifying the

disease in its early stages, as measurement of the intima-media complex allows suspicion already in phase I of the pathology, when intima-media thickening occurs because of inflammatory infiltrate [8]. Treatment includes systemic corticosteroid therapy, since there is an active inflammatory process, and the association with immunosuppressive agents may be necessary. Surgical approaches may also be required, depending on the affected vessel and the complications it causes, such as arterial grafting with venous substitutes or even angioplasty with stent placement as a second option [8].

This case report aims to discuss a 28-year-old patient with Down syndrome (DS) diagnosed with TA, addressing the diagnostic pathway and raising the possibility of an association through immunogenetic interactions between trisomy 21 and vascular diseases such as TA. Furthermore, there is a bibliographic gap clarifying this relationship, and no case reports associating DS and TA were identified, which reinforces the relevance of this case report in exploring a possible etiopathogenic link.

2. Case Report

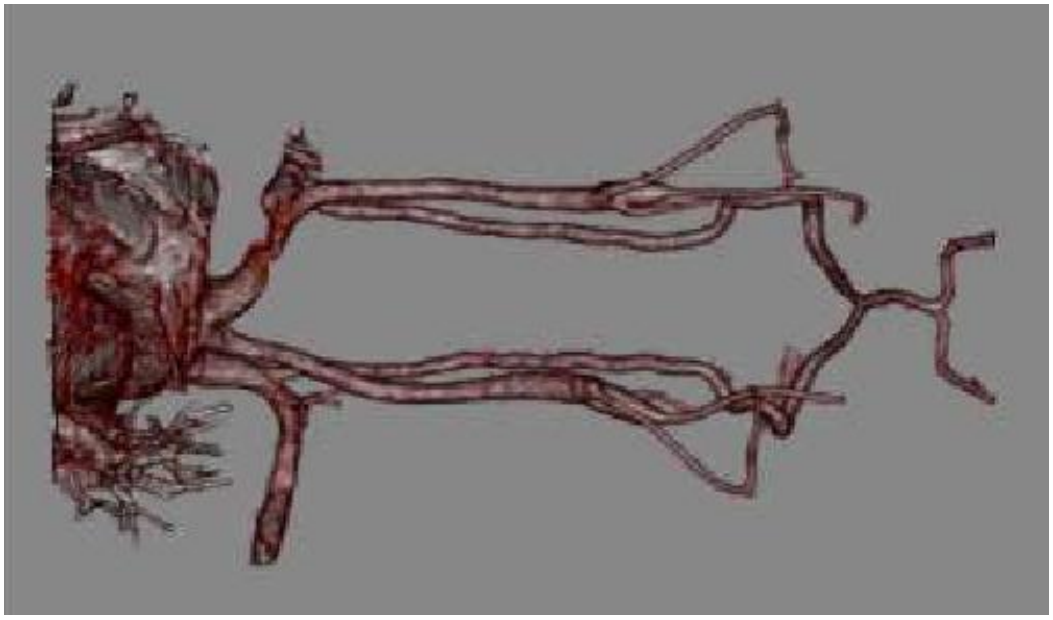
A 28-year-old white female patient with Down syndrome (DS) was admitted to the emergency care unit presenting with dysarthria and right-sided hemiplegia, with symptom onset 48 hours prior to admission. On directed medical history, a previous history of upper limb claudication during usual physical activities was reported, present for several months before the current neurological event. She also described recurrent early postprandial gastric fullness, even after small meals, suggestive of mesenteric angina. These symptoms preceded the acute neurological condition and were not systematically investigated. There were no reported fever, documented weight loss, or clear constitutional symptoms, although this information was limited due to reliance on family reports. The patient denied smoking, alcohol consumption, and illicit drug use. She had no known comorbidities and was regularly using acetylsalicylic acid, atorvastatin, and omeprazole.

On initial physical examination, she was conscious, with a Glasgow Coma Scale score of 15, presenting difficulty in speech articulation. A blood pressure difference of 60 mmHg between the upper limbs was observed, with measurements of 180×100 mmHg in the right arm and 120×100 mmHg in the left arm. On auscultation, bruits were identified bilaterally over the carotid arteries and over the right subclavian artery. Peripheral pulses showed asymmetric reduction, and neurological examination revealed right hemiparesis, consistent with cerebral ischemia. There were no signs of heart failure, significant abdominal abnormalities, or other relevant systemic findings.

Laboratory tests demonstrated marked systemic inflammatory activity, with significant elevation of erythrocyte sedimentation rate (ESR ranging from 102 to 110 mm/h between April 19 and 25, 2024) and C-reactive protein (CRP), which peaked at 59.9 mg/L on April 19, 2024, remaining persistently elevated in subsequent measurements. These findings reinforce the presence of active inflammatory disease. Coagulation tests performed during the same period did not reveal significant coagulopathy (INR 1.08). Other laboratory tests, including complete blood count and comprehensive metabolic panel, were within normal limits.

Additional diagnostic tests were performed, including cranial computed tomography (CT), which demonstrated areas of deep hypodensity in the left corona radiata and, to a lesser extent, on the right side, as well as hypodensity of the deep white matter in the right parietal lobe, findings consistent with cerebral ischemia. Cervical CT angiography demonstrated diffuse abrupt narrowing of the bilateral internal carotid arteries, beginning just above the carotid bifurcations (proximal thirds) and extending to the terminal intracranial portions, without evidence of collateral circulation. No abnormalities of the subclavian arteries were described (Figure 1). Thoracic and abdominal CT angiography revealed stenosis in the mid-segment of the brachiocephalic trunk, without significant involvement of the abdominal aorta or renal arteries. Tissue biopsy was not feasible.

Figure 1. Coronal reconstruction computed tomography showing a hypodense formation (arrowhead), predominantly cystic, located below the right hepatic lobe and extending to the cecal region.



Based on the combination of clinical, laboratory, and imaging findings, the diagnosis of Takayasu arteritis was established. The patient was treated with intravenous corticosteroid pulse therapy, followed by systemic corticosteroid therapy, showing significant clinical improvement, with remission of abdominal claudication, improvement of pulse asymmetry, and regression of neurological deficits, including dysarthria and hemiparesis. After clinical stabilization, the patient was discharged with recommendation for specialized outpatient follow-up. However, despite active attempts to contact the patient, she was lost to follow-up, and it was not possible to document inflammatory laboratory progression or long-term adherence to immunosuppressive therapy.

3. Discussion

Takayasu arteritis is a progressive disease and can be characterized as a transmural granulomatous vasculitis mediated by T lymphocytes; studies have also demonstrated that genetic components are involved in the pathogenesis of Takayasu arteritis [9]. It may cause stenosis, aneurysm formation, and occlusion of affected vessels. It is a primary systemic vasculitis with a pattern of fibrinoid necrosis. This case report is relevant because it addresses the occurrence of TA in a young female patient with Down syndrome. The literature describes abnormal vascular morphology in individuals with trisomy 21; therefore, we suggest that the coexistence of these two pathological entities may not be coincidental.

The clinical manifestations of TA depend on the phase in which the disease presents. Typically, as in the reported case, the presentation is sudden or subacute, which, combined with the absence of a specific disease marker, delays appropriate diagnosis. An important point is that the reported patient presented TA in the third phase, as she exhibited predominant signs of inflammatory exacerbation that progressed to stenosis and fibrosis, resulting in an ischemic condition [10]. Neurological manifestations occur in more than half of patients during the course of the disease, among which stroke is one of the most severe complications. When stroke constitutes the initial manifestation of the disease, it tends to cause significant delay in diagnosis and treatment initiation, with direct impact on prognosis [11].

The diagnosis of TA follows the 2022 American College of Rheumatology (ACR) criteria, which include clinical and imaging parameters (Table 1). In our case report, the total score was 11 points, confirming the diagnosis [12,13]. However, in this case, although there was no involvement of the subclavian arteries, the patient fulfilled the diagnostic criteria proposed by the American College of Rheumatology. Subsequently, according to the 2021 criteria [14], a concomitant diagnosis of TA and Moyamoya syndrome was proposed, also referred to as quasi-moyamoya disease, due to the association of Down syndrome with steno-occlusive involvement of the terminal portion of the internal carotid artery, but without the development of Moyamoya vessels.

Table 1. Classification Criteria for Takayasu Arteritis According to the American College of Rheumatology (2022).

Criteria	Variables	Score
Absolute requirement	Age ≤ 60 years at the time of diagnosis	—
	Imaging evidence of vasculitis [1]	—
Additional clinical criteria	Female sex	+1
	Angina or ischemic cardiac pain	+2
	Claudication in leg or arm	+2
	Vascular bruit [2]	+2
	Reduced pulse in upper extremities [3]	+2
	Carotid artery abnormality [4]	+2
	Systolic blood pressure difference between arms ≥ 20 mmHg	+1
Additional imaging criteria	Number of affected arterial territories (select one) [5]:	
	One arterial territory	+1
	Two arterial territories	+2
	Three or more arterial territories	+3
	Symmetric involvement of paired arteries [6]	+1
	Abdominal aorta involvement with renal or mesenteric involvement [7]	+3

* Add the score for the 10 items if present. A total score of ≥ 5 points is required for classification as Takayasu arteritis.

Notes: Evidence of vasculitis in the aorta or its main branches must be confirmed by vascular imaging, such as computed tomography, catheter-based angiography, magnetic resonance angiography, ultrasound, or positron emission tomography. A bruit refers to a sound detected by auscultation over a large artery, including the aorta, carotid, subclavian, axillary, brachial, renal, or iliac arteries. Reduced or absent pulse on physical examination may involve the axillary, brachial, or radial arteries. It may also refer to reduced or absent carotid artery pulse or carotid artery tenderness. The number of arterial territories with luminal damage, such as stenosis, occlusion, or aneurysm, detected by angiography or ultrasound includes the following major territories: thoracic aorta, abdominal aorta, mesenteric arteries, left or right carotid arteries, left or right subclavian arteries, and left or right renal arteries. Bilateral luminal damage (stenosis, occlusion, or aneurysm) detected by angiography or ultrasound may occur in any of the following paired arterial territories: carotid, subclavian, or renal arteries. Luminal damage may also involve the abdominal aorta and/or the renal or mesenteric arteries, as detected by angiography or ultrasound. Source: The final 2022 American College of Rheumatology/EULAR classification criteria for Takayasu arteritis [12].

It is described in the literature that individuals with Down syndrome (DS) are genetically predisposed to developing certain vascular conditions, with inheritance patterns showing variable expressivity [15–17]. Proteins encoded on chromosome 21 have an impact on arterial physiology and elasticity, although this association has been more strongly addressed in the literature in relation to cardiac pathologies [16]. These proteins are related to oxidative stress, including superoxide dismutase-1, increased interferon-gamma pathway activity, and cystathionine synthase [16]. There is also overexpression of type VI collagen, whose alpha-chain protein is encoded on chromosome 21 and is found

in the lining of large arteries, which may consequently result in malformed blood vessels [17,18].

Comparatively, in TA there is production of cytokines such as interferon-gamma (IFN-γ), whose serum level may reflect disease activity [19]. Variants in the gene encoding type V collagen have also been identified [20], as well as increased markers of oxidative stress [21]. Some of the main vascular and circulatory conditions associated with Down syndrome include congenital heart disease, pulmonary hypertension, peripheral vascular disorders, autoimmune and inflammatory diseases, as well as premature aging and atherosclerosis [17].

In a study involving two genotyped cohorts of patients with TA, an additional genetic association effect conferring risk for TA was detected on chromosome 21 (21q22, the critical region for DS), with a p-value < 0.05, representing a modest but genomically significant association, particularly among European-American individuals [9]. This finding requires further studies to ensure that it was not due to chance. The most significant genetic association detected was in the HLA region. Despite shared mechanisms, the absence of previous reports of TA in patients with DS and the lack of targeted genomic studies preclude definitive conclusions.

Although individuals with DS have an increased risk of vasculitis, the low frequency of cases associated with TA raises the hypothesis of underdiagnosis or even misclassification with other occlusive vasculopathies, such as Moyamoya syndrome (MMS). Isolated diagnosis of TA is rare, and several authors attribute this low detection rate to reduced clinical awareness of the disease in Western countries compared to Eastern regions, which substantially contributes to delayed diagnosis. The cerebrovascular involvement cluster of TA, with its ongoing active inflammation, may exacerbate cerebral artery stenosis, similarly to MMS [22]. The association between MMS and DS is widely reported in the literature, supporting the hypothesis that DS exhibits variable phenotypic expressivity within the spectrum of vascular involvement.

MMS, characterized by progressive narrowing of the internal carotid artery and consequent increased risk of ischemic cerebrovascular events, provides a plausible model of how intrinsic vascular alterations in Down syndrome may culminate in large-vessel disease [17]. Recent studies demonstrate that the RNF213 gene, in interaction with chromosome 21, has been described as promoting mutations that influence vascular physiology and elasticity in patients with DS, potentially resulting in the previously described MMS manifestations. Variants of the RNF213 gene have also been associated with other intracranial and extracranial vasculopathies independently of MMS, and there is evidence suggesting that it may represent a susceptibility gene for TA, being mainly related to angiogenesis and arterial remodeling. Deficiency or mutation leads to impaired vascular patterning and increased fragility, contributing to steno-occlusive pathology [23]. Some studies propose that this gene may act as a unifying factor in vasculopathies, and murine studies are currently underway. It is also believed that epigenetic factors may influence which vascular phenotype will manifest. In this case, exome sequencing, if available, for genetic and pathogenic evaluation of the reported patient, could assist in clarifying the relationship between DS and TA.

The cornerstone of TA treatment is high-dose corticosteroid therapy (1–2 mg/kg/day) during the active phase of the disease for a period of one to three months [24]. In critical phases, pulse therapy (500–1,000 mg for 1 to 3 days) may be used [25]. It is also possible to associate other immunosuppressive drugs, such as methotrexate (20–25 mg weekly) or azathioprine (2 mg/kg/day), as they improve relapse and remission rates [26,27]. It should be emphasized that there are no clinical studies specifically involving patients with TA that allow the establishment of a standardized treatment model. In the present case, after pulse therapy with hydrocortisone, the patient showed satisfactory clinical evolution, allowing hospital discharge.

Regarding prognosis, it is estimated that in patients who do not develop disease complications (retinopathy, hypertension, aortic insufficiency, or aortic aneurysm), survival is

approximately 90%, whereas in those who develop complications, survival is approximately 60% at 15 years [26]. However, these data may not be extrapolated to patients with DS, as there are no studies correlating these two conditions.

4. Conclusion

Thus, the reason for the association between TA and DS is not yet fully understood, and several hypotheses have been proposed. We suggest a possible immunogenetic predisposition associated with the interaction between the *RNF213* gene and genes located on chromosome 21, influencing vascular physiology and anatomy, as observed in Moyamoya syndrome. Further research is necessary to confirm or refute this association. It is imperative that, in patients with Down syndrome who present symptoms such as pulse deficits, claudication, and angina, the diagnosis of Takayasu arteritis be considered in order to establish early diagnosis and more appropriate treatment.

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