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CASE REPORT

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Cerebrofacial arteriovenous metameric syndrome type I + II + III: An unusual case

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Key Clinical Message

Cerebrofacial arteriovenous Metameric syndrome (CAMS) typically manifests as types I, II, or III, occasionally presenting as dual types. Our unique case underscores the coexistence of all three CAMS types in one patient. Furthermore, the concurrent acute cerebellar infarct underscores the need to consider CAMS in the differential diagnosis of adolescents experiencing neurological events.

KEYWORDS

arteriovenous malformations, CAMS, cerebral infarct, hydrocephalus, seizures, VP shunt

1 | INTRODUCTION

Cerebrofacial arteriovenous metameric syndrome (CAMS), also recognized as cerebrofacial vascular metameric syndrome, Wyburn–Mason Syndrome, and Bonnet–Dechaume–Blanc Syndrome, represents a complex disorder characterized by metameric distribution involving retinal, facial, cerebral, and cerebellar vascular malformations.[1] It is classified into three subgroups based on prosencephalon or rhombencephalon involvement. CAMS I encompasses the medial prosencephalic group involving the hypothalamus, pituitary, and nasal regions. CAMS II involves the lateral prosencephalic group, which includes the occipital lobe, thalamus, optic tract, retina, and maxilla. CAMS III encompasses the lateral rhombencephalon group involving the cerebellum, nasal bridge, and mandible. The pathophysiology of CAMS remains poorly understood, with speculation suggesting that somatic mutations occurring in neural crest cells immediately before migration may lead to the metameric lesions observed in CAMS.[2] We present a case of CAMS type I + II + III:

2 | CASE HISTORY

An 18-year-old male presented to the emergency department accompanied by his parents, reporting a one-day history of recurrent generalized tonic–clonic seizures. Additionally, the patient had experienced dizziness, vertigo, and a tendency to fall on the left side over the past week. Previous consultations with a local health practitioner resulted in the prescription of vitamin supplements without a comprehensive workup. Further inquiry revealed a progressive decline in visual acuity, memory loss, and recurrent epistaxis over the past 8 years, with a similar seizure episode occurring 1.5 years ago and no drug abuse history.

Upon examination, the conscious and oriented patient exhibited vitals within normal ranges but presented with facial asymmetry, proptosis of the left eye, and cutaneous lesions on the left side of the face and scalp. Notably, the patient displayed positive cerebellar signs, including abnormalities in the finger-nose and heel–knee tests on the left side. A lean physique with a short stature was also observed.

3 | METHODS

Basic metabolic panel results were unremarkable, prompting a plain CT brain to rule out intracerebral hemorrhage or space-occupying lesions, revealing hydrocephalus and abnormally dilated vascular channels along the orbit, brainstem, and cerebellum.

Prophylactic anticonvulsants were prescribed, and subsequent MRI brain and orbits identified an intracranial nidus of dilated vascular channels predominantly affecting the left thalamus, hypothalamus, brainstem, fourth ventricle, and cerebellum, causing mass effects and hydrocephalus, and the patient underwent ventriculoperitoneal shunting (VP shunt) for hydrocephalus. A concurrent extracranial nidus was observed along the left scalp and face. An acute infarction in the left cerebellar hemisphere was also evident on imaging. Cerebral angiography was performed for a three-dimensional assessment of the disease extent.

4 | CONCLUSION AND RESULTS

Upon confirmation of CAMS (Figure 1–7), the patient was discharged and referred for radiosurgery treatment at a specialized neurology center. Subsequent follow-up indicated conservative management without intervention, emphasizing regular observations and evaluations.

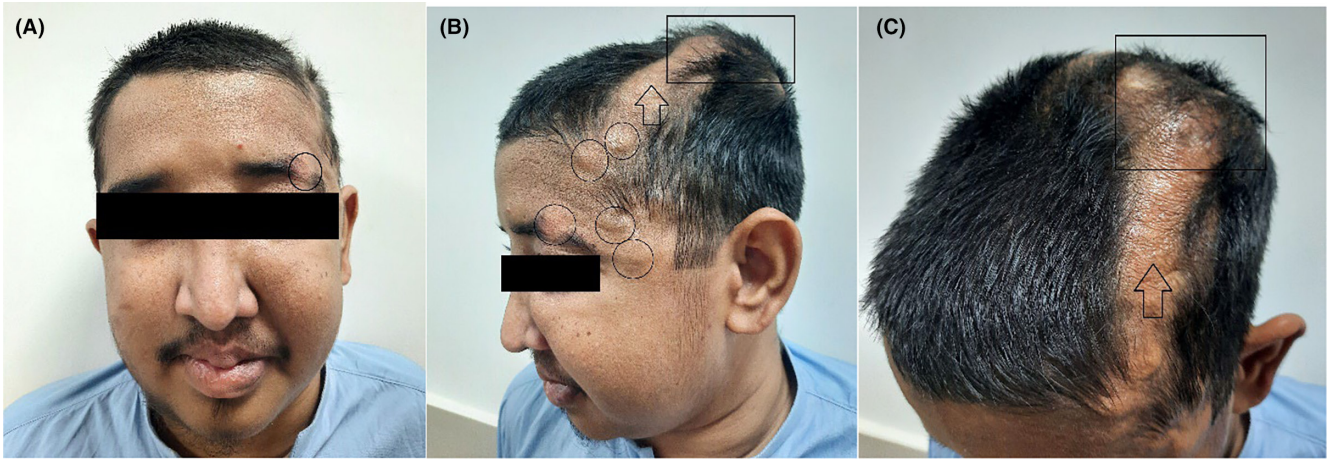

FIGURE 1 Gross images of the patient demonstrate facial asymmetry, tortuous cutaneous lesions (black circles), loss of scalp hair in left temporoparietal region (black arrows) and raised soft tissue swelling in left superior parietal region (black box).

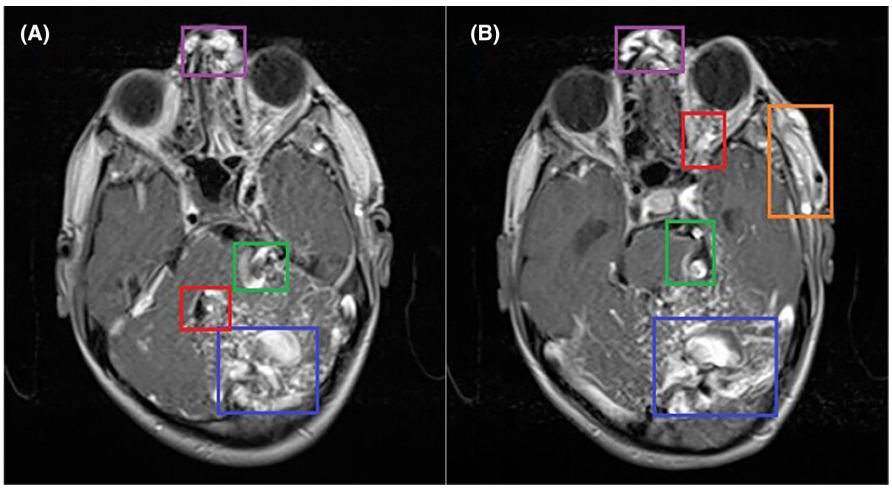

FIGURE 2 MRI brain and orbits. (A) Axial T1-weighted image with contrast enhancement demonstrates arteriovenous malformation (AVM) involving the left cerebellar hemisphere (blue box), the fourth ventricle (red box), the left cerebellopontine angle (green box) and the external nose (purple box). (B) Axial T1-weighted image with contrast enhancement demonstrates AVM involving the external nose (purple box), left maxilla and soft tissue (orange box), the left retrobulbar space causing mild proptosis (red box), the basilar cisterns (green box) and both cerebellar hemispheres, more on the left side (blue box).

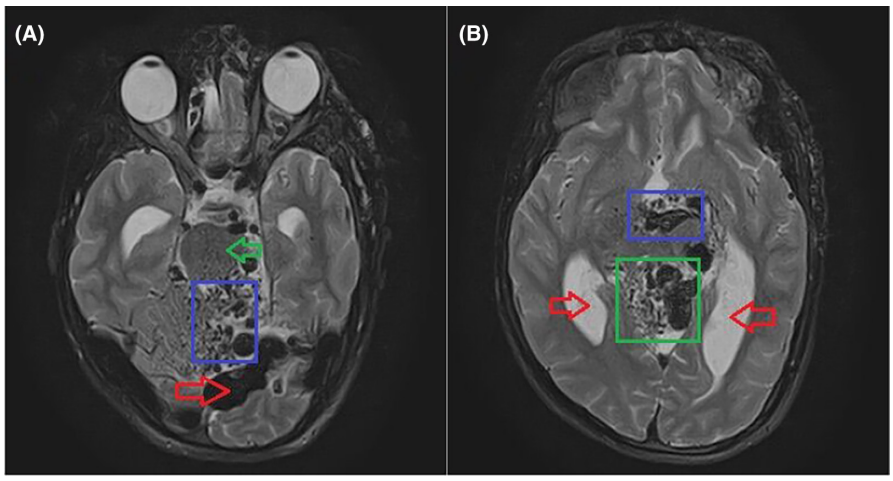

FIGURE 3 MRI brain and orbits. (A) Axial T2-weighted fat-saturated image demonstrates AVM (hypointense indicating signal void) involving the left cerebellar hemisphere (blue box), causing a mass effect on the pons displacing it to the right (green arrow). A dilated draining vein is also seen (red arrow). (B) Axial T2-weighted fat-saturated image demonstrates AVM involving hypothalamic region (blue box) and basal cisterns (green box), causing obstructive hydrocephalus and leading to dilation of the occipital horns of the lateral ventricles (red arrows).

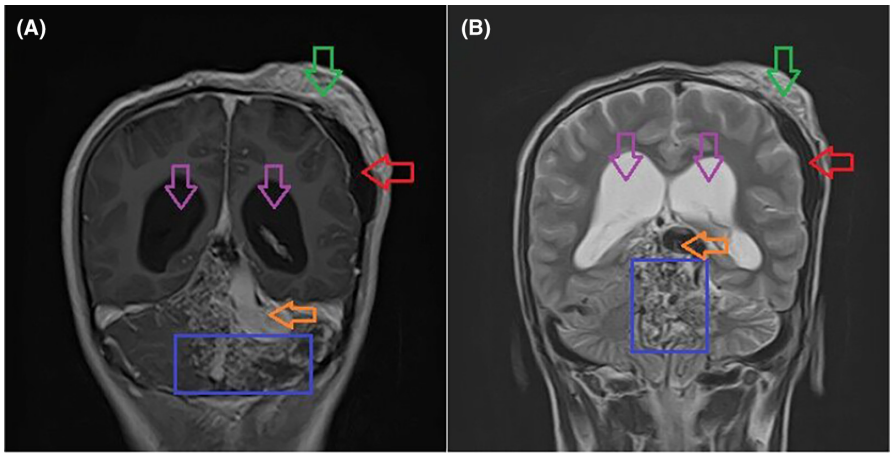

FIGURE 4 MRI brain and orbits. (A) Coronal T1-weighted image with contrast enhancement and (B) Coronal T2-weighted image with contrast enhancement demonstrate AVM involving the cerebellar hemispheres (blue box) and the left superior parietal region causing a soft tissue swelling (green arrow) and hyperostosis of the underlying parietal skull bone (red arrow). Dilation of the lateral ventricles (purple arrows) and dilation of a draining vein (orange arrow) is also seen.

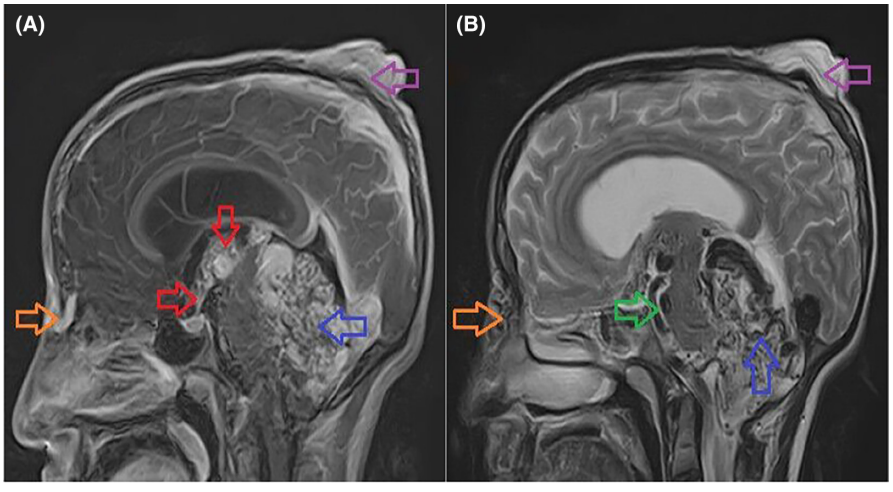

FIGURE 5 MRI brain and orbits. (A) Sagittal T1-weighted image with contrast enhancement and (B) Sagittal T2-weighted image demonstrates AVM involving cerebellar hemispheres (blue arrow), brainstem (green arrow), thalamic and hypothalamic areas (red arrows), superior parietal region (purple arrow) and the external nose (orange arrow).

5 | DISCUSSION

Cerebrofacial arteriovenous metameric syndrome is a rare, non-inherited condition characterized by an arteriovenous malformation affecting the brain, orbit, and face. Bonnet, Dechaume, and Blanc reported the first documented association of this malformation within these anatomical regions in 1937.[3] Bhattacharya later proposed a metameric nature for the condition, attributing it to a disorder in neural crest development. Consequently, he introduced the term cerebrofacial arteriovenous metameric syndrome (CAMS) to describe this unique medical phenomenon.[2]

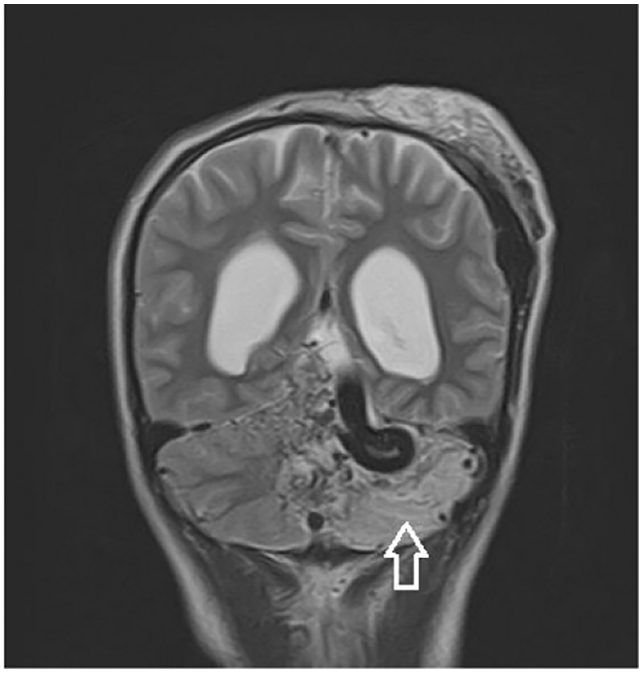

FIGURE 6 MRI brain coronal T2-weighted demonstrates hyperintense area in the left cerebellar hemisphere (white arrow) suggesting an acute infarction.

Cerebrofacial arteriovenous metameric syndrome is a disease that affects people of all ages but usually presents in childhood. Clinical presentations range from weakness, red eyes, epistaxis, and facial discoloration to CSF rhinorrhea, gait ataxia, seizures, headaches, and angiomatous lesions. Gait ataxia and CSF rhinorrhea are uncommon presentations that indicate neurological complications associated with CAMS.[4–12] Imaging techniques include a protocol consisting of CT brain, CT angiography, and MRI brain. Arteriovenous malformations were identified in different anatomical regions, including the Willis polygon, orbit, suprasellar region, brainstem, cerebellum, thalamus, hypothalamus, and optic nerve. CAMS types I, II, and III were diagnosed based on the location and extent of vascular malformations. CAMS type II is the most common malformation type, especially in cases with reported orbital and temporal regions.

Our case stands out as it satisfies the criteria for all three types of CAMS, a presentation never before seen in the literature. The findings of our case are consistent with CAMS types I, II, and III, and this case is the first of its kind. The rarity of this case, as highlighted by the

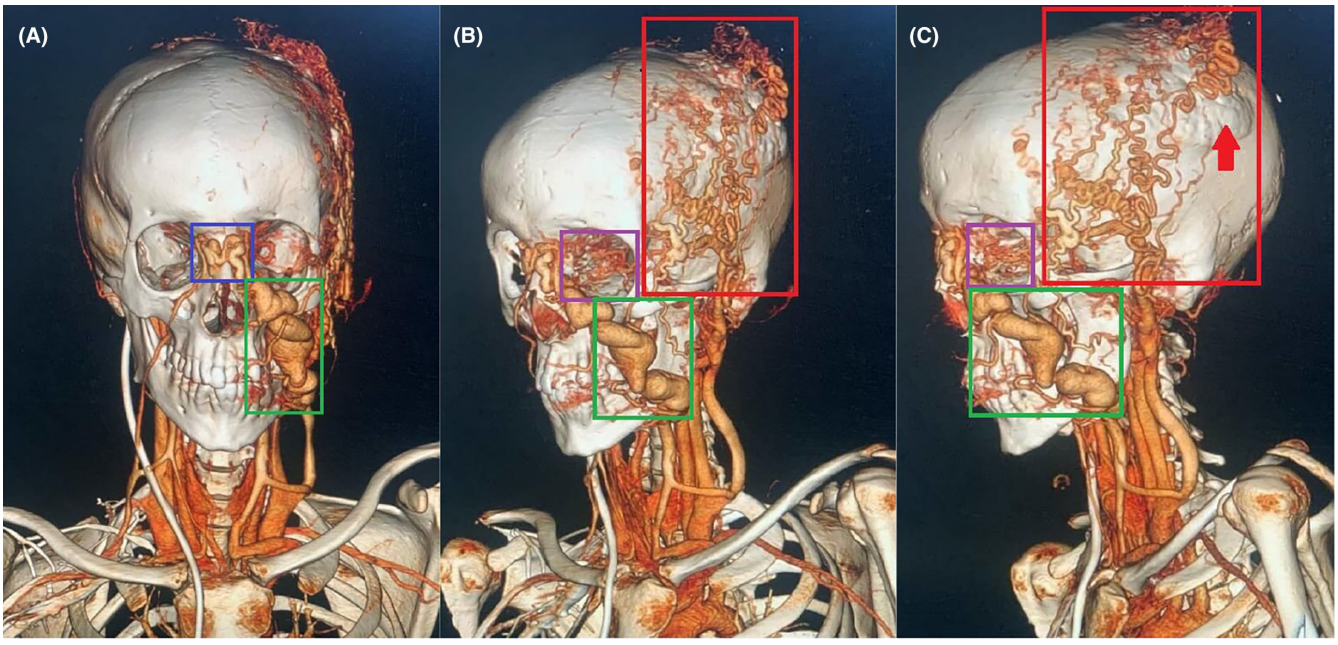

FIGURE 7 CT cerebral angiography (A–C) demonstrates AVM involving the external nose (blue box), the left orbit (purple box), the maxillary and mandibular region (green box) and the left temporoparietal region (red box) with hyperostosis of the underlying parietal skull bone (red arrow).

TABLE 1 Summarizes some of the reported cases of CAMS.

Authors	Title	Age & gender	Geographical location	Diagnostic technique	Type of CAMS
da Silva, et al 2021[4]	Cerebrofacial vascular metameric syndrome associated with Moyamoya syndrome: a rare case report	7 months old, female	NA	MRI T2 weighted and cerebral angiography	Type II
Vasanthapuram, et al 2020[5]	Cerebrofacial arteriovenous metameric syndrome type 2	3 year old, male	India	An MRI (T2-weighted) imaging with contrast revealed a distinct and varied enhancing hyperintense lesion located in the left superior ophthalmic fissure and posterior orbit. Additionally, there is an observed prominence of the left sphenoparietal sinus and pterygoid venous plexus, along with multiple venous anomalies detected in both thalami and cerebellar hemispheres on both side	Type II
Ng, et al 2018[6]	Cerebrofacial arteriovenous metameric syndrome with hypopituitarism: a rare association	9 year old, male	NA	Series of MRI at age 2, 5, and 8 years and four-vessel angiogram. Fundoscopy shows dilated retinal vessels on the right side. and anterior nasal endoscopy shows prominent vessels on the little's area and inferior turbinate on the right.	Type I and II
Fernández-Gajardo, et al 2018[7]	Ethmoid Meningoencephalocele in a Patient with Cerebrofacial arteriovenous metameric syndrome	45 year old, female	NA	Magnetic resonance imaging (MRI) and digital subtraction angiography revealed the presence of an arteriovenous malformation (AVM) on the right side affecting the face and orbit, with an extension reaching posteriorly along the optic tract into the suprasellar cistern.	Type II
O'Loughlin, et al 2017[8]	Cerebrofacial arteriovenous metameric syndrome (CAMS): a spectrum disorder of craniofacial vascular malformations	9 year old female and 14 year old female	NA	Diagnosis involves utilizing MRI and confirming through CT angiography, which enables the delineation of angioarchitecture and real-time assessment of hemodynamics in lesions.	Type II
Kang, et al 2005[9]	Cerebellopontomandibular vascular malformation: a rare type of cerebrofacial arteriovenous metameric syndrome. Case report	22 year old, female	NA	Magnetic resonance imaging detected widespread absence of vascular signals across the entire cerebellar hemisphere, vermis, and right-sided pons. Angiography revealed cerebellar, facial and pontine AVMs on the right side	Type III

(Continues)

TABLE 1 (Continued)

Authors	Title	Age & gender	Geographical location	Diagnostic technique	Type of CAMS
Haw, et al, 2003[11]	Coexistence of mandibular arteriovenous malformation and Cerebellar arteriovenous malformation An Example of Cerebrofacial arteriovenous metameric syndrome type III	6 year old, female	NA	CT scan mandible and cerebral angiography was done	Type III
Jiarakongmun, et al, 2002[10]	Clinical course and angioarchitecture of cerebrofacial arteriovenous metameric syndromes. Three demonstrative cases and literature review	28 year old, male. 4 year old, male. 6 year old, male	NA	MRI and cereberal angiogram was used as imaging modality in all three case	NA

limited number of case reports in the existing literature (Table 1), presents a challenge in diagnosing and treating this condition. The aberrant vascular formations within specific regions may lead to disturbances in blood flow, resulting in ischemic events and infarcts that could explain the acute infarction in left cerebellar hemisphere in this case. Moreover, the presence of dilated vasculature could predispose individuals to thromboembolic events by promoting stasis. Treatment modalities and protocols need time, especially because most of the cases in the literature have only mentioned conservative treatment of the disorder due to the location of the lesions.[6] Further collaboration is required between neurologists, radiologists, and other specialized medical personnel for effective management protocols for patients suffering from CAMS.

Our case highlights the atypical presentation of the CAMS, revealing the three common types (I, II, and III) in a single patient. Recognizing this unique variability in CAMS manifestations is essential for accurate diagnosis and appropriate management. The rarity of these cases emphasizes the importance of fostering a stronger awareness among medical professionals, as this complexity may not be consistent with traditional diagnosis expectations. Furthermore, the inclusion of acute cerebellar infarction in this case further emphasizes the importance of taking CAMS into account in the differential diagnosis of adolescents with brain infarction. CAMS, with its different clinical manifestations, can lead to complications such as infarction, which may be overlooked if not specifically considered in the diagnostic process. This global understanding is essential for timely interventions and appropriate management strategies. Recognizing that CAMS can represent a combination of types, health care providers can enhance their diagnostic abilities to ensure that patients receive the most effective and targeted care.

AUTHOR CONTRIBUTIONS

Shahroze Ahmed: Data curation; investigation; resources. **Digbijay kunwar:** Conceptualization; data curation; methodology; writing – original draft. **Muhammad Hamza Khan:** Methodology; writing – original draft; writing – review and editing. **Anum Akbar:** Conceptualization; methodology; project administration; supervision; writing – original draft; writing – review and editing. **Asra Yasin:** Data curation; investigation. **Abdul Sattar Anjum:** Data curation; investigation; resources.

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None.

CONFLICT OF INTEREST STATEMENT

None.

DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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