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Case Study of Atypical KlippelTrenaunay Syndrome Radiology

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ABSTRACT

Aim: To explore the various radiological findings associated with Klippeltrenaunay syndrome in this unilateral lower limb involvement using colour Doppler sonography, contrast enhanced CT, and MR venography. By employing these imaging modalities, we hope to identify both common and unusual features of the condition and emphasize the importance of CT and MR venography in complementing colour Doppler sonography to fully assess the extent of the syndrome's involvement.

Case details

- A 19 year old male came to the OPD with complaints of externally visible multiple superficial tortuosities in right lowerlimb predominantly on antero-lateral aspect of thigh since birth and formation of superficial cutaneous ulcerations on leg since 1 month
- **On examination**: A prominent tortuous vein is seen coursing upwards from right thigh through right iliac fossa and horizontally crossing the midline towards left groin.
- Another prominent tortuous vein is seen coursing upwards on the right lateral abdominal wall.
- Multiple ulcerations noted on the medial aspect of right leg.

Conclusion: Our case study demonstrates common and unusual findings of KlipperTrenaunay Syndrome and highlights the role of CT and MR venography in addition to color Doppler sonography to identify the complete extent of the involvement.

Keywords: KlippelTrenaunay syndrome, Imaging, Lower limbs, Deep venous aplasia

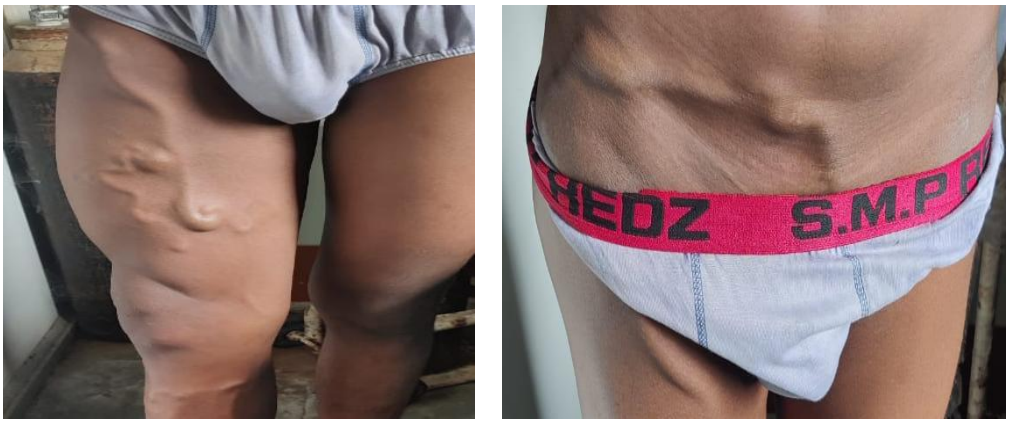
INTRODUCTION

- Klippeltrenaunay syndrome is an uncommon congenital disorder [1] distinguished by a trio of manifestations, namely vascular malformations or port-wine stains, varicose veins, and excessive growth of either bony or soft tissue.Diagnosis of this syndrome requires the presence of atleast two of these features, with capillary malformations being the most common. However, atypical klippeltrenaunay syndrome can also be diagnosed in patients without capillary malformations.
- Notable differential diagnoses includesParkes Weber syndrome, Beckwith-Wiedemann syndrome, Maffucci syndrome and macrodystrophialipomatosis [2].Some cases of Klippeltrenaunay Syndrome have exhibited deep venous abnormalities, including aplasia, duplications, and aneurysmal dilatation [2]. The identification of these deep venous malformations is crucial for understanding the extent of The lateral marginal vein of Servelle and sciatic vein are the two embryonic veins usually seen persistent in KlippelTrenaunay Syndrome [3].
- While conventional venography used to be the gold standard, it has been largely replaced by noninvasive modalities like colour Doppler, CT, and MR venography due to their lower invasiveness.The presence of varicosities in unusual locations such as lateral aspect of the leg, and deep venous anomalies are highly indicative of Klippel-Trenaunay syndrome.
- In this condition,Vascular (venous) malformations commonly manifest as a complex arrangement of interconnected blood vessels, accompanied by a mixture of soft tissue and the presence of calcified phleboliths.

- Spectral Doppler, examination reveals low flow in vascular malformations, exhibiting no flow or monophasic flow. KlippelTrenaunay syndrome is typically characterized by absence of high flow arterio-venous malformations[4].
- A plain radiograph of the affected limb can potentially reveal the existence of phleboliths or intra-osseous extension associated with the malformations.
- For preoperative mapping of the venous system, CT or MR venography can be employed to identify the extent of deep seated malformation and help prevent untoward events during interventions on superficial varicosities. Due to the absence of ionizing radiation, MRI is often preferred as the primary modality, particularly for younger patients, making it the favored choice [4].
- CT venography offers advantages over MR venography, including shorter procedure time and higher spatial resolution. However, the management of KTS is primarily conservative[5-7], focusing on symptomatic relief, addressing bleeding episodes, and preventing and treating complications like deep vein thrombosis, cellulitis, chronic coagulopathy, and congestive heart failure.

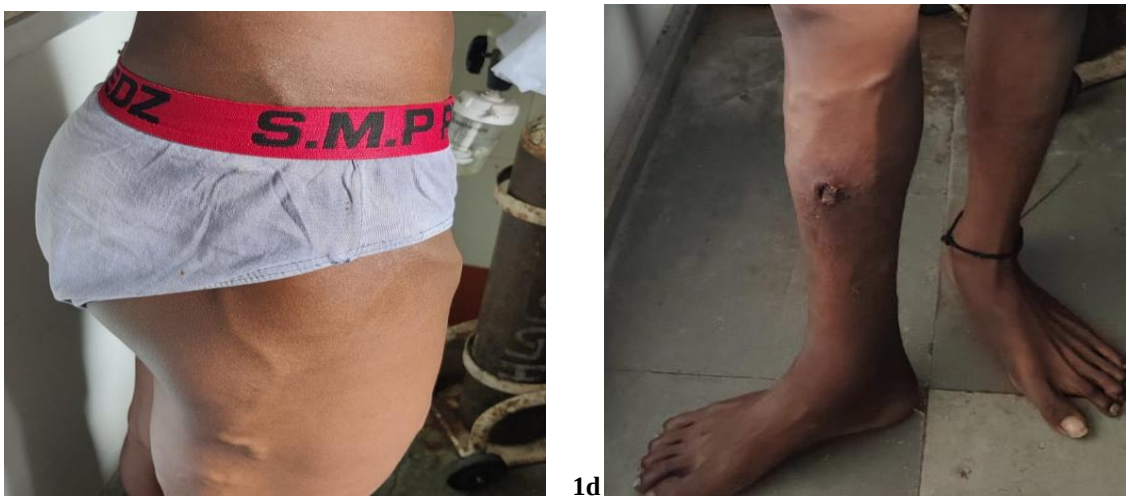
CASE PRESENTATION

A 19-year-old male patient presented with a non-healing ulcer in the right leg since 1 month with varicosities in the right lower limb, which were present since childhood. He was being managed conservatively for the varices.



1a **1b**

Images 1a&1b: Clinical photographs of patient are showing hypertrophy of right lower limb muscles with multiple externally visible tortuous varices in right thigh and right leg predominantly on anterolateral aspect.A prominent vein is seen coursing horizontally from right iliac fossa towards left groin seen in image 1b.



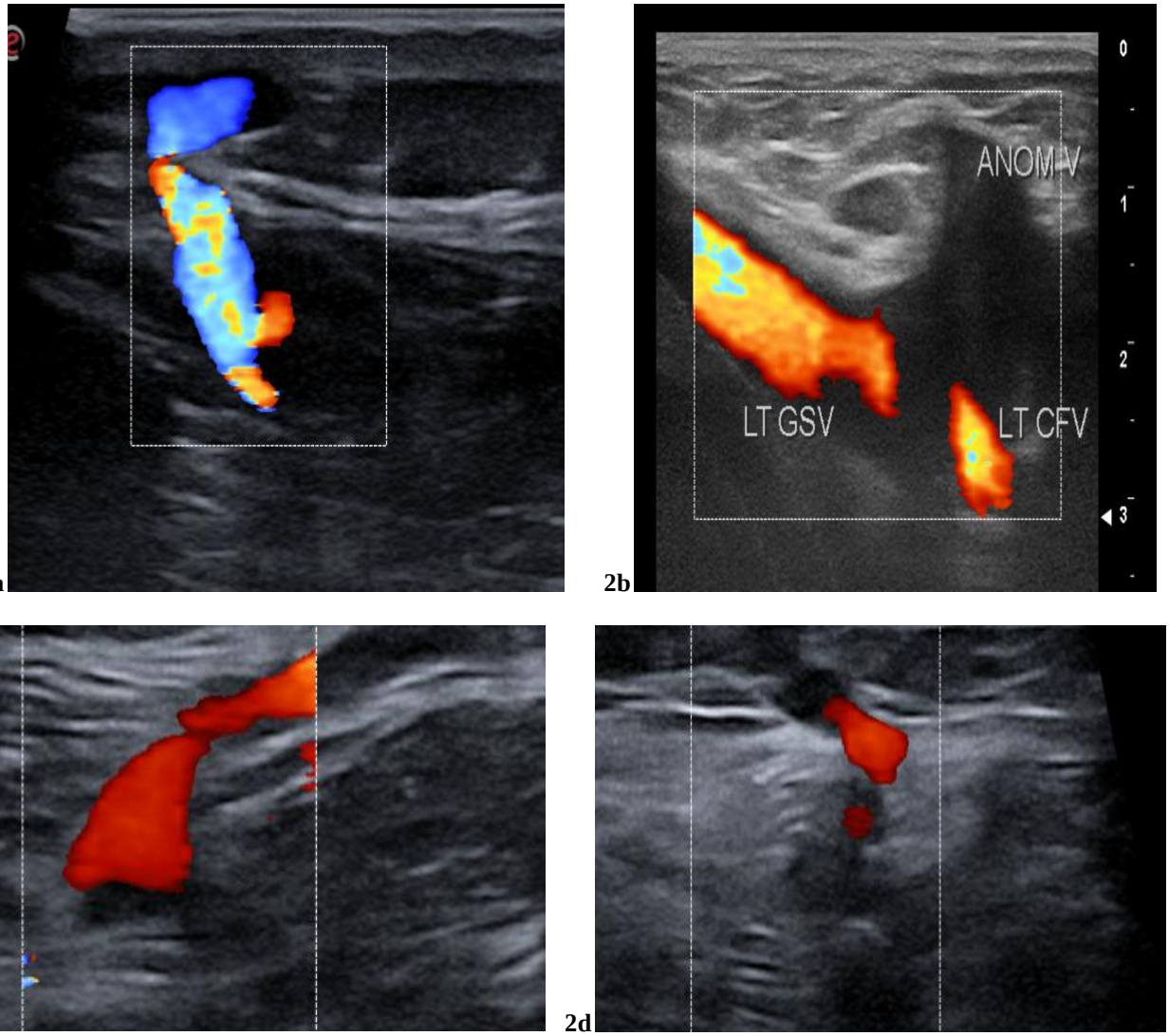
1c **1d**

Images 1c&1d: Clinical photographs of patient are showing aprominent tortuous vein coursing upwards on the right lateral abdominal wall. There is a cutaneous ulcer on medial aspect of middle 1/3rd of hypertrophied right leg seen in image 1d.

On external examination:

- Multiple varicosities are noted in hypertrophied right lower limb predominantly in antero-lateral aspect. A prominent tortuous vein is seen coursing upwards from right thigh through right iliac fossa and horizontally crossing the midline towards left groin. (Fig.1a& 1b)
- Another prominent tortuous vein is seen coursing upwards on the right lateral abdominal wall(Fig. 1c).
- Multiple ulcerations noted on the medial aspect of right leg (Fig. 1d).

The patient was scheduled for surgical treatment of the varicose veins, and as part of the preoperative assessment; a color Doppler evaluation study of varicose veins was arranged (Mylab X6 Esoate Ultrasound machine). Doppler study revealed extensive varicosities involving ananomalous venous channelwhich drains the right lower limb. Scanning of the deep venous system showed a complete absence of right external iliac, common and superficial femoral, profundafemoris & popliteal veins. Suspecting a congenital agenesis of deep venous system, a CT venography was performed using a multi-detector row 16-slice CT scanner (General electronics 16 slice scanner) which confirmed the Doppler findings of complete absence of deep venous system of the right lower limb in addition to hypertrophied right lower limb, extensive varicosities along the course of anomalous dilated venous channel which takes an unusual anterolateral ascent in right thigh through right iliac fossa & further vertical ascent until it drains into infra-renal IVC on right lateral aspect. One branch from the anomalous vein taking a horizontal course through the midline from right iliac fossa region towards left groin & finally drains into left common femoral vein.(Fig. 2). Another branch from anomalous venous channel travels in anterolateral aspect of abdominal wall ascending towards azygos vein & draining into it [Fig 3,4]. MR Venography confirmed the CT venography findings with great soft tissue resolution [Fig. 3-5].

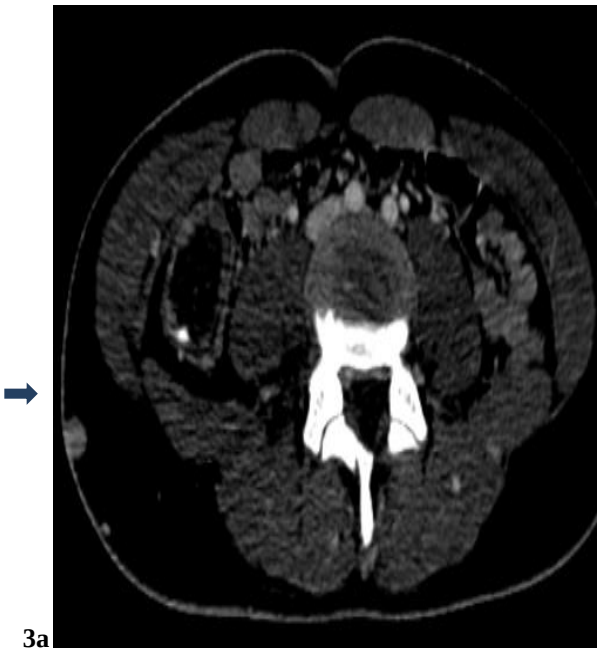


2a 2b

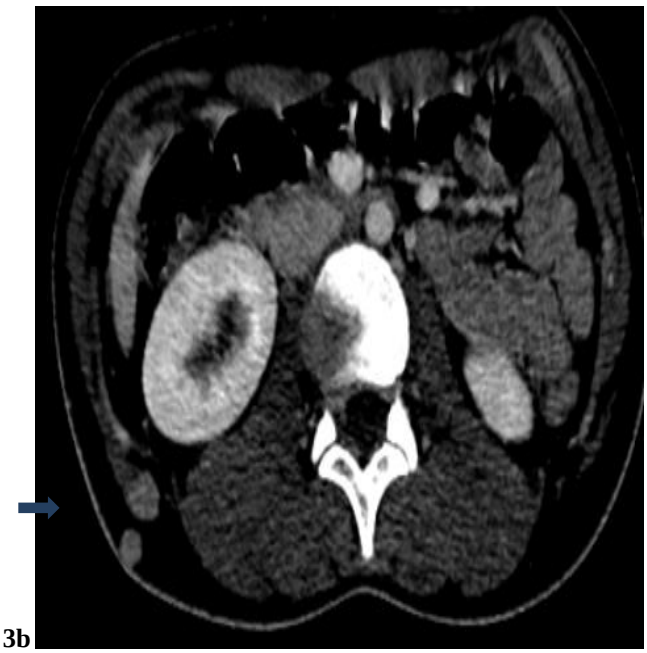
2c 2d

Fig 2a,b,c,d: Color doppler findings include normal deep venous system noted on left side

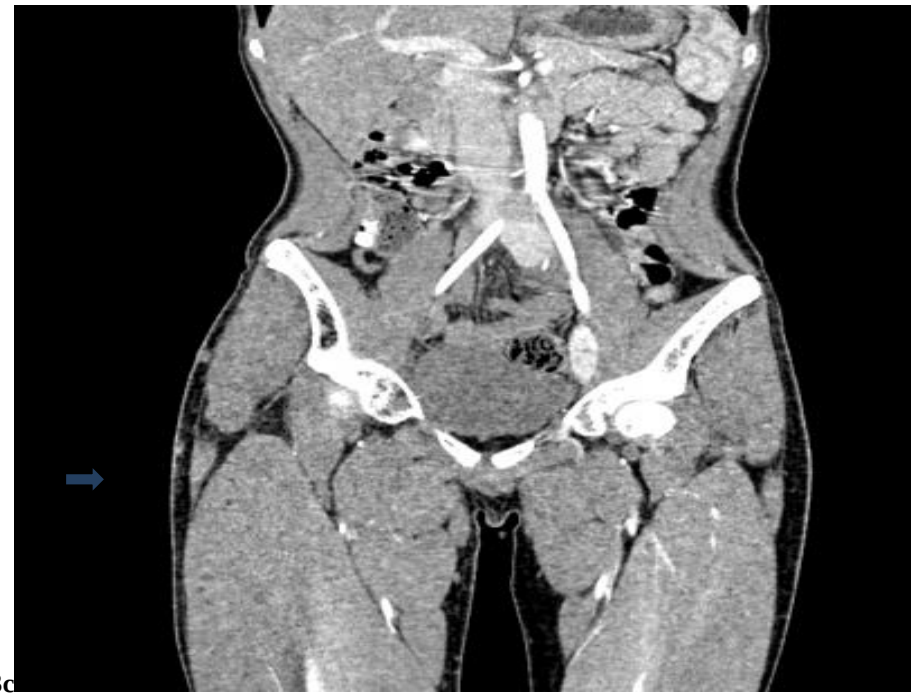
Complete Absence of deep venous system on right side
Anomalous tortuous vein is seen crossing midline from right side and joining into left common femoral vein



3a



3b



3c

Image 3a.b.c: CECT findings include normal IVC caliber. IVC is drained by single left common iliac vein. Left lower limb deep & superficial venous systems appear normal.Complete absence of right common, internal and external iliac veins and deep femoral system. Multiple superficial anomalous veins noted in right lower limb predominantly in non- saphenous distribution.

The arrow marked vein when traced upwards is seen draining into the azygos vein
Lower limb muscles on the right side appear hypertrophied when compared to left side


A branch from long anomalous tortuous venous channel is seen arising at the right groin going superiorly, crossing midline and draining into left common femoral vein

Fig 4

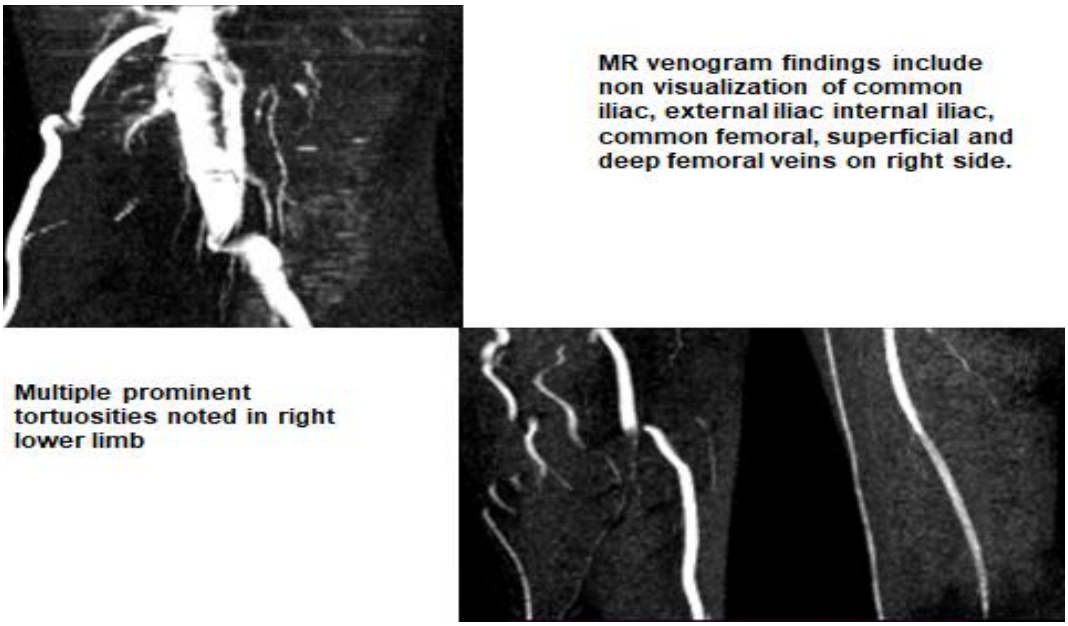

MR venogram findings include non visualization of common iliac, external iliac internal iliac, common femoral, superficial and deep femoral veins on right side.
Multiple prominent tortuosities noted in right lower limb

Fig 5


2 long prominent superficial venous channels carrying venous drainage from right lower limb are seen, one of them draining into left common femoral vein and the other one into azygos vein

Fig 6

DISCUSSION

KTS is a syndrome characterized by extremely variable clinical features and severity of symptoms as well as remarkable variability in the expression of its components. Over the years, various authors have utilized different diagnostic criteria for this entity. Evolution of advanced imagingtechniques definitelyincreased the ability of detection and characterization of the various vascular malformations;

A strict diagnostic criterion proposed by Oduber et al. emphasizes the variable nature of KlippelTrenaunay syndrome (KTS) [8]. As per the criterion, the diagnosis of KlippelTrenaunay Syndrome (KTS) requires the presence of two primary diagnostic features: (a) congenital vascular malformation and (b) abnormal growth of the affected limb. Vascular malformations in KTS are classified as capillary malformation (port-wine stain), venous malformation, arterio-venous malformation, and lymphatic malformation. However, the presence of capillary and venous malformations is crucial for diagnosing KTS. Venous malformation serves as a broad term encompassing varicose veins, vein hypoplasia or aplasia, and persistent fetal veins. The growth disturbance in KTS involves hypertrophy or hypotrophy of small or large body parts, ranging from isolated finger involvement to entire limb or half body-involvement. Our study patient satisfied the diagnostic criteria for KTS. Hypertrophy, as observed in our case is the typical feature of KTS. Additionally, other imaging findings, though not specific to KTS, have been observed in some cases, including hemimegalencephaly, hemangioma, aneurysm, autonomic dysfunction, cavernoma, gastrointestinal and genitourinary vascular anomalies, polydactyly/syndactyly, and positional limb defects like talipesequinovarus and or metatarsus varus [8,10].

The evaluation of KTS severity primarily focuses on assessing the clinical severity of venous abnormalities, encompassing the extent and distribution of structural and functional venous abnormalities.

Comprehensive Doppler sonography allows for a thorough assessment of the affected limb, providing indication of the severity of venous insufficiency in the superficial venous system. Furthermore, evaluating the perforator system and deep venous system aids in classifying the extent of venous reflux as either triple (involving superficial, deep, and perforator veins) or dual (involving superficial veins with either deep or perforator veins) venous insufficiency. Additionally, factors such as deep venous hypoplasia, deep venous thrombosis, limb edema, leg ulcers, superficial thrombophlebitis, and intra-osseous extension of vascular malformation, arthritis, and neuropathy contribute to the overall morbidity. Although our case study did not involve a quantitative pulsed wave Doppler evaluation of venous insufficiency, it did indicate a higher severity of the disease [11].

In conclusion, our case study highlighted the diverse imaging findings observed in KlippelTrenaunay Syndrome affecting the lower limbs. Diagnosis of KTS requires a high index of suspicion, especially when there are notable imaging findings such as a lifelong history of symptoms, unilateral limb hypertrophy, and an atypical distribution of varices that may extend into the muscles and bones. Evaluation of the superficial venous system should focus on identifying the unusual distribution of varicosities associated with low flow venous malformations and recognizing persistent embryological veins like the lateral marginal vein. Furthermore, careful assessment of the deep venous system is important to identify any partial or complete agenesis or duplication. Our case study also highlights the importance of obtaining supplementary imaging with CT or preferably MR venography to fully evaluate the extent of the disease, particularly when it may involve the underlying bones or organs.

Abbreviations CT: Computed tomography; KTS: Klippel-Trenaunay syndrome; MRI: Magnetic resonance imaging
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