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# Pelvico-Abdominal Plexiform Neurofibromatosis: Radiologic Clues to an Exceptional Presentation

**Regina Primeylisa Dos Santos[1]*, Pande Putu Yuli Anandasari[2]**
Rumah Sakit Umum Pusat Prof. dr. I.G.N.G. Ngoerah, Indonesia
Email: regina.dsantos31@gmail.com*, yulianandasari@unud.ac.id

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**ABSTRACT**

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder with a prevalence of approximately 1 in 4,000–5,000 individuals worldwide. Plexiform neurofibroma, a less common but clinically significant subtype, occurs almost exclusively in NF1 patients and carries a lifetime risk of malignant transformation to malignant peripheral nerve sheath tumors (MPNSTs) of approximately 4.6%–13%. *Pelvico-abdominal* plexiform neurofibromas are exceptionally rare, with fewer than 60 cases reported in the literature. This study aims to describe a unique case of *pelvico-abdominal*plexiform neurofibromatosis in an adolescent female and to highlight the essential role of multimodality imaging—including ultrasound, CT, and MRI—in establishing the diagnosis, assessing anatomical involvement, detecting complications, and guiding clinical management. This is a case report of a 14-year-old female patient presenting with progressive abdominal enlargement over three years. Diagnostic evaluation included plain abdominal radiography, abdominal ultrasound, contrast-enhanced CT, MRI with gadolinium, and histopathological examination of a retroperitoneal biopsy. Imaging revealed a large, heterogeneous solid mass extending from the pelvic cavity to the abdominal cavity. Ultrasound demonstrated the characteristic "target sign" appearance, with hyperechoic centers and hypoechoic peripheries, appearing hypovascular on Doppler imaging. CT showed a low-attenuation heterogeneous mass displacing adjacent organs (rectum, colon, uterus, and bladder) and encasing major vessels (abdominal aorta, iliac arteries, and inferior mesenteric artery) without evidence of invasion. MRI confirmed a confluent, multinodular mass with multiple target signs on T2-weighted images and central enhancement on contrast-enhanced T1-weighted Dixon sequences. Histopathology confirmed plexiform neurofibroma with no mitotic activity.

## INTRODUCTION

Neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF2) are distinct genetic conditions characterized by the development of noncancerous tumors known as neurofibromas (Peduto et al., 2023; Moodley et al., 2024). Although there are some similarities between the two, they exhibit different clinical characteristics and genetic compositions (Griborio-Guzman et al., 2022; Taylor et al., 2023; Wang et al., 2022). The disease is caused by a mutation in the NF1 gene located on chromosome 17q11.2 and is inherited in an autosomal dominant manner (Liao et al., 2025; Ga o et al., 2022). The prevalence of NF1 is approximately 1 in 4,000–5,000, making it one of the most common hereditary multilocus tumor syndromes (Coşkun et al., 2020; Moodley et al., 2024). The main features of NF1 involve the central nervous system

(CNS), skin, and bones, with clinical manifestations including osseous lesions such as sphenoid dysplasia, axillary or inguinal freckling, neurofibromas or plexiform neurofibromas, optic nerve gliomas, Lisch nodules, and café-au-lait macules (Liao et al., 2025; Santangelo et al., 2025; Zhang, 2024).

A number of additional manifestations of NF1 include low-grade gliomas, vascular dysplasia, various abdominopelvic neoplasms, and interstitial lung disease (Ferner et al., 2021; Gutmann et al., 2020). NF2 is commonly associated with tumors such as ependymomas, meningiomas, and schwannomas (Ruggieri & Huson, 2021; Evans et al., 2022). NF1 can be classified into two subtypes: localized and plexiform. Plexiform neurofibromas are diffuse, locally invasive tumor masses that extend along nerve sheaths, grow around distorted nerve bundles, and can spread along adjacent nerves, muscles, and skin (Wagner et al., 2024; Ratner & Miller, 2020). These lesions may grow superficially or deeply and are associated with high morbidity due to their progressive growth and potential for tissue damage. These tumors carry a significant risk of malignant transformation, particularly within the nervous system (Friedman et al., 2022). Pelvico-abdominal neurofibroma is a rare condition, with approximately 60 cases reported in the literature (Smith et al., 2023). We report a unique case of pelvico-abdominal neurofibromatosis in an adolescent female and highlight the importance of multimodality imaging in its diagnosis (Khayat et al., 2021).

The urgency of this research stems from several factors. First, the rarity of pelvico-abdominal plexiform neurofibromas means that clinicians may not consider this diagnosis when encountering a large abdominal mass in an adolescent, potentially leading to diagnostic delays or unnecessary invasive procedures. Second, the significant risk of malignant transformation (approximately 20-fold increased risk) necessitates accurate diagnosis and appropriate surveillance. Third, the complex anatomical location involving pelvic organs, major vessels, and the urinary and gastrointestinal tracts requires precise preoperative imaging for surgical planning. Fourth, the adolescent patient population presents unique considerations regarding growth, development, fertility, and long-term cancer risk that are not fully addressed in adult-focused literature. Therefore, this case report is needed to raise clinical awareness and provide practical imaging guidance.

The novelty of this research lies in several key aspects. First, this case represents one of the few documented instances of pelvico-abdominal plexiform neurofibroma in an adolescent female, adding to the limited literature on this exceptional presentation. Second, the study provides a comprehensive multimodality imaging correlation—including plain radiography, ultrasound with Doppler, contrast-enhanced CT, and MRI with gadolinium—from a single patient, offering a unique opportunity to compare the diagnostic utility of each modality. Third, the research highlights specific imaging clues (e.g., the “target sign” on ultrasound and MRI, low attenuation on CT, and vascular encasement without infiltration) that can aid radiologists in differentiating plexiform neurofibroma from other pelvico-abdominal masses such as rhabdomyosarcoma, paraganglioma, ganglioneuroma, and leiomyosarcoma. Fourth, the study discusses the clinical implications of these imaging findings for management decisions, including surgical planning and surveillance for malignant transformation.

The purpose of this research is to describe a unique case of pelvico-abdominal plexiform neurofibromatosis in an adolescent female and to demonstrate the essential role of multimodality imaging in diagnosis. The research contribution is twofold: clinically, it

provides a practical imaging guide for radiologists encountering similar cases; educationally, it raises awareness among clinicians about this rare but important differential diagnosis. The objectives are to: (1) present the clinical, imaging, and histopathological findings of this exceptional case; (2) correlate the imaging features with known pathological characteristics of plexiform neurofibromas; (3) compare the diagnostic utility of different imaging modalities; and (4) discuss management implications based on imaging findings. The benefits of this research include improving diagnostic accuracy, facilitating appropriate patient counseling regarding malignant transformation risk, guiding surgical planning, and contributing to the limited body of literature on pelvico-abdominal plexiform neurofibromas in the adolescent population.

## METHOD

A 14-year-old female patient came with a complaint of an enlarged stomach since 3 years ago, an enlarged stomach accompanied by a palpable lump on the abdomen. Initially, the patient did not realize that there was a lump in the abdomen until finally the patient's stomach felt enlarged. The size of the lump on the abdomen has been the same since 3 years ago and there is no pain in the abdomen or lump in the abdomen. The history of nausea and vomiting is denied. Other complaints were denied, previous trauma history was denied. Patients daily activities as students and and lumps do not interfere with the patient's activities

On physical examination, it was found that the abdomen was enlarged accompanied by a lump on almost the entire abdomen, palpable, there was no pain when palpation. Laboratory examination, complete blood test, blood electrolyte examination and blood gas analysis within normal limits.

From the results of AP abdominal polo photos, it was found that there was opacity in the pelvic cavum to the abdominal cavum.

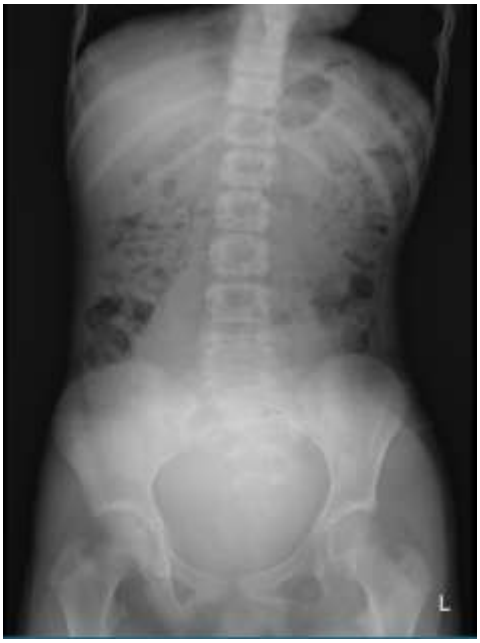


Figure 1. Plain photo of AP's abdomen shows opacity in the pelvic cavum to the abdominal cavum.

The results of the abdominal ultrasound examination showed the presence of heterogeneous solid masses in the pelvic cavity to the abdominal cavity accompanied by "target-signs" with hyperechoic center and hypoechoic edges that in the doppler appear hypovascularized.

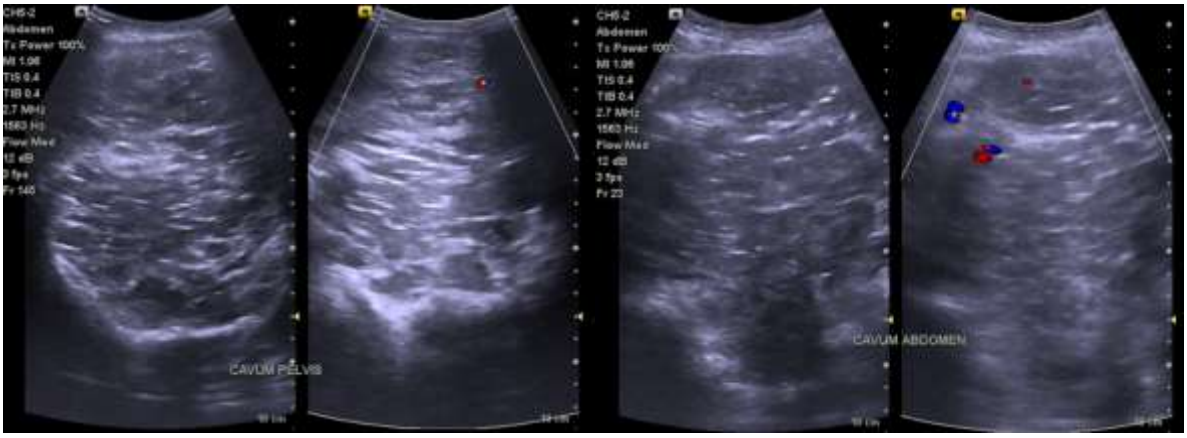


Figure 2. Abdominal ultrasound examination showed the presence of heterogeneous solid masses in the pelvic cavity to the abdominal cavity accompanied by "target-signs" with hyperechoic centers and hypoechoic edges that appear hypovascular in Doppler.

On CT Scan, a picture of a heterogeneous solid mass with low density in the pelvic cavum extends to the abdominal cavum, which appears to push the rectum and colon distal to the anterior, urges and presses the uterus to the anterocranial, urges and presses the bulge to the right anterolateral, urges the intestinal system to the cranial and peripheral, the mass also appears to be clouding the abdominalis aorta, a. illiaca communis right left, a. illiaca externa and internal right left, and encapsulated a. inferior mesenterica, the mass boundary with the surrounding organs is still good, there is no infiltration and the tk also appears to be extended to the bone.

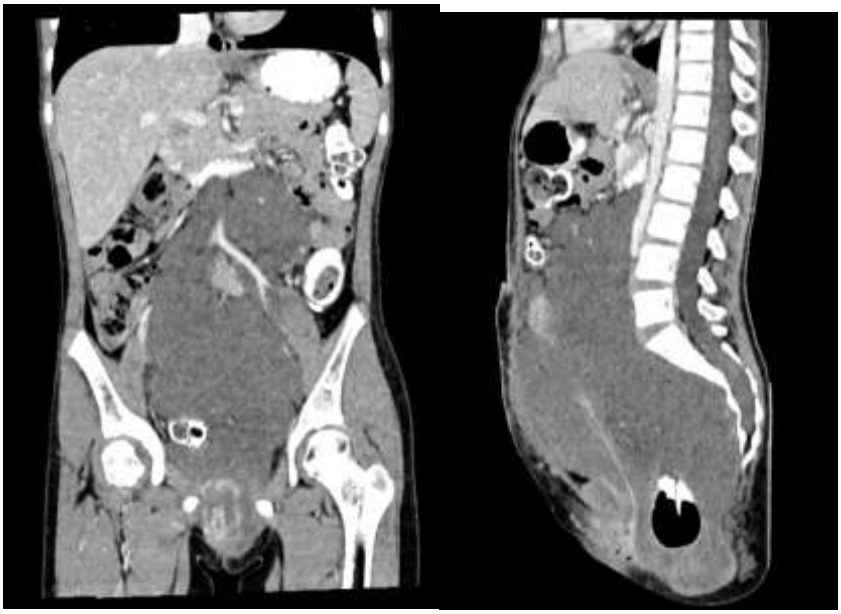

Figure 3. CT Scan examination obtained A picture of a heterogeneous solid mass with a low density in the pelvic cavity extending to the abdominal cavity

Meanwhile, the examination of the abdomen with contrast obtained a wide hyperintense multinodular confluent mass with several "target-sign appearance" appearances with hypointense signal intensity in the clear central part and hyperintense signal intensity in the peripheral part of the T2 Haste as well as an increase in intensity in the central part in the T1 Dixon W with gadolinium contrast.

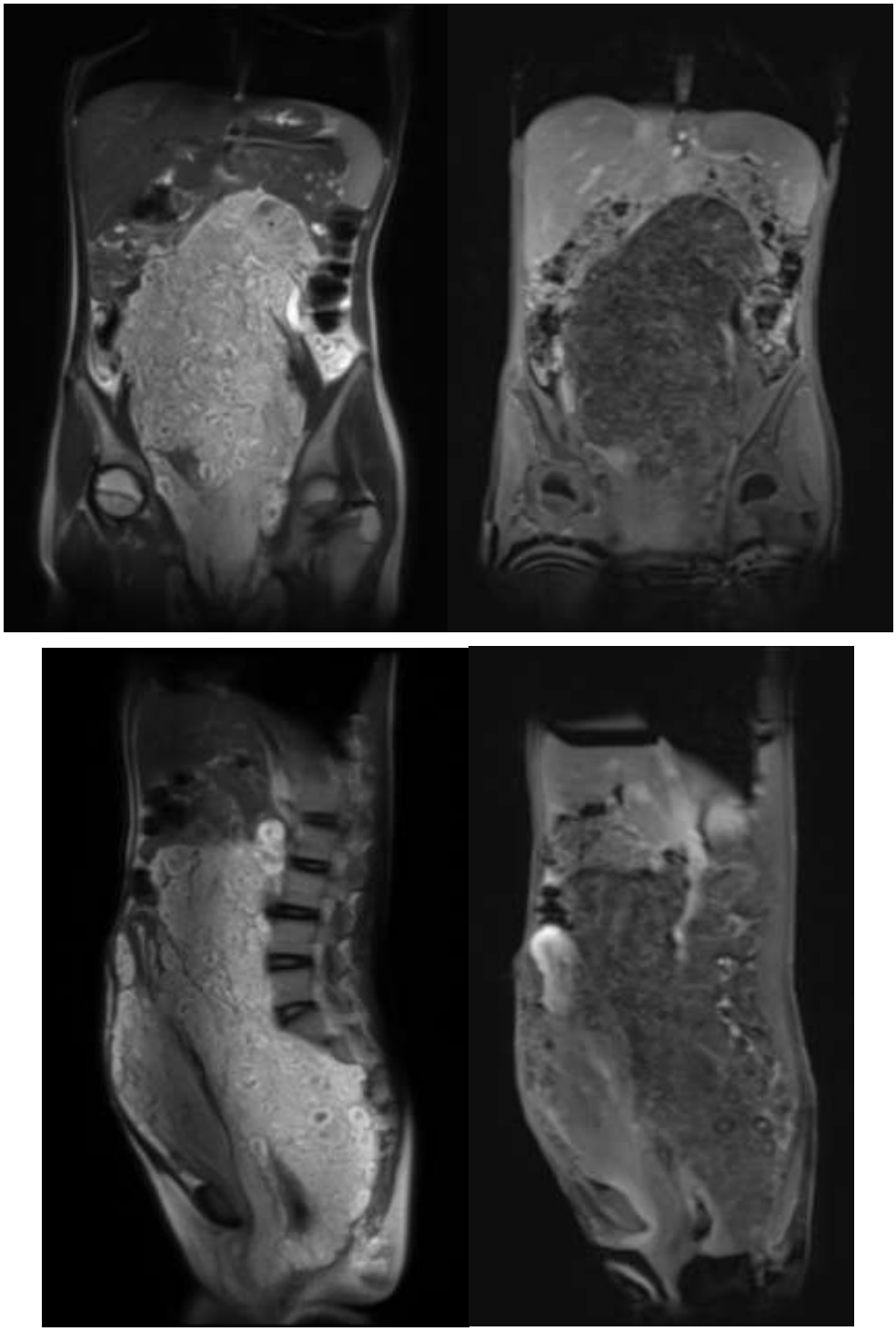

Figure 4. MRI examination of the coronal & sagittal fragment showed a broad hyperintense confluent multinodular mass with multiple images of "target sign appearance" with intensity of hipointens in the mid and hyperintense in the peripheral part in T2 Haste and central enhancement in T1 Dixon W with contrast

Histopathology of retroperitoneal biopsy contains a mass consisting of fibrocollagen fibers arranged in a loose arrangement, among which it appears that myxoid material contains cells with spindle-shaped and ovoid cell nuclei and lymphocyte aggregate focuses. Mitosis was not found.

## RESULT AND DISCUSSION

Neurofibromas are neoplasms that originate from Schwann cells, are located in any part of the body, and are mostly found on the skin. Neurofibromatosis is a hereditary condition that can be identified by certain diagnostic criteria, such as the presence of one or more neurofibromas, Lisch nodules in the iris, some café-au-lait spots, bone changes, optic nerve gliomas, and other symptoms. Plexiform neurofibroma, a less common type of benign neurofibroma, is a tumor that occurs specifically in individuals with NF I. These tumors occur due to uncontrolled growth of all nerve components in the peripheral nerves. The term "plexiform" describes the nucleus of interconnected or intertwined networks of blood vessels or nerves, as observed in this particular condition. The term "plexiform" comes from the Latin verb "plex," which signifies the act of weaving or weaving.

Pelvic neurofibromas can appear either independently or as a component of a multiorgan disorder known as NF-1. Although rare, malignant degeneration can occur. Diagnosing pelvic-abdominal neurofibroma is easier when. associated with other syndromes. However, in isolated cases, an appeal diagnosis for masses at the base of the bladder should include rhabdomyosarcoma, paraganglioma, ganglioneuroma, and leiomyosarcoma.

Plexiform neurofibromas appear as thickened, hard masses or nodules that can infiltrate structures, causing deformation and dysfunction. On MRI, they manifest as multinodular confluent masses with a mass effect on the surrounding structures and some target marks[1] that have the potential to transform into malignant peripheral nerve sheath tumors (MPNST). MPNST has a poor prognosis due to metastasis to the brain, lungs, liver, soft tissues, bones, regional lymph nodes, skin, and retroperitoneum. The lifetime risk of neurofibromatosis undergoing malignant transformation is estimated to be 4.6%[8]. Therefore, patients with plexiform neurofibromas are 20 times more likely to develop MPNST compared to patients without plexiform neurofibroma

We perform a comprehensive assessment of our patients using ultrasound (ultrasound), CT Scan and MRI. Imaging is important for a variety of reasons: it helps assess the level of involvement and its impact on nearby structures, detects associated abnormalities, and, most importantly, malignant transformations. Ultrasound is often performed to assess the mass of palpable soft tissue. Differential diagnosis depends on the characteristics of the tumor components. Peripheral nerve sheath tumors can exhibit a variety of sonography features, with most appearing as well-defined oval-shaped hypooecoic masses connected to peripheral nerves.

The mass often shows a target-like appearance on ultrasound, characterized by a hyperechoic center and darker outer edges with increased posterior elevation behind it. On color Doppler imaging (CDU), these structures usually appear to have reduced blood flow, although there may be some cases where internal flow is observed[10]. It is very important to distinguish these tumors from vascular malformations and infections such as abscesses.

Vascular malformations can be ruled out with CDU, since the tumor does not show increased blood flow during compression and release. The tumor does not have echoes of internal movements, which is characteristic of abscesses. Nonetheless, it is important to recognize that ultrasound does not have the ability to consistently distinguish between benign and malignant lesions.

Neurofibromatosis is characterized by a mass of soft tissue with low attenuation on a CT scan. This is due to the presence of myelin-lipid content, high water content, and fats trapped in the endoneurial myxoid tissue. [11]Our patients showed similar low-attenuation lesions in the intraperitoneal and extraperitoneal regions, with no bone involvement. CT scans provide an advantage in assessing bone involvement. In suspected malignant lesions, CT scans show heterogeneity with visible necrosis in the central part, unclear borders, and irregular peripheral nodular increases.

MRI is still the best way to diagnose neurofibromatosis (NF) with the characteristics of "target marks" on T2-weighted images, hyperintense peripheral edges and hypointense central fibrous components, as well as central enhancement. In the case of plexiform neurofibroma, MRI shows a multinodular confluent mass with multiple target marks and mass effects on nearby structures. It is essential to get a definitive soft tissue diagnosis to rule out malignancy, as plexxiform neurofibromas have the potential to become malignant. Since the target sign is absent in some neurofibromas, it is not a reliable indicator of malignancy. MRI is essential to distinguish between neurofibroma and Malignant Peripheral Nerve Sheat Tumor (MPNST). This is done by evaluating the dimensions of the increased largest mass, swelling around the lesion, the presence of fluid-filled sacs inside the tumor, and peripheral increase patterns. The presence of 2 to 4 of these features indicates malignancy, with specificity and sensitivity of 90% and 61%, respectively.

Treatment of plexiform neurofibromas requires a multidisciplinary approach, usually involving surgery, that aims to remove deformed masses and cancerous tissue as malignant transformations occur. In addition, complete removal is often difficult due to extensive tumor growth, invasion of surrounding tissue, and the tendency to regrow after surgery. In some studies in patients who did not have surgery, it was possible to take medications such as interferon and mitogen-activated protein kinase (MEKi), such as selumetinib, which may reduce the volume and symptoms caused by the mass. Taking these factors into account and avoiding ionizing radiation, MRI is the best modality for patient monitoring. The most effective primary treatment of neurofibromatosis malignant transformation is complete surgical resection. Additional therapies, such as radiotherapy or neoadjuvant therapy, including chemotherapy, are options for cases that cannot be resected or to reduce the risk of recurrence. However, the prognosis remains unpredictable due to the high risk of developing the disease and its varied manifestations.

## CONCLUSION

Plexiform neurofibroma is a characteristic nerve sheath tumor most commonly associated with NF1, distinguished by multilobulated masses and the classic “target sign” on imaging. These tumors can involve both superficial tissues and deeper anatomical structures, reflecting their infiltrative growth along nerve pathways. On ultrasound, CT, and MRI, they typically demonstrate mixed fat and fluid components related to myelin content, making

radiologic evaluation crucial for accurate diagnosis, assessment of anatomical extent, differentiation from other pathologies, and early detection of malignant transformation to guide management decisions. Future research should focus on developing advanced imaging biomarkers and longitudinal studies to improve early detection of malignant change and to better stratify patient risk, particularly in pediatric and adolescent populations.

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