Journal of Biosciences and Medicines, 2026, 14(1), 15-27
https://www.scirp.org/journal/jbm
ISSN Online: 2327-509X
ISSN Print: 2327-5081

Application of Imaging Modalities in the Diagnosis of Branchial Anomalies

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How to cite this paper: Chen, Y.W. and He, X.M. (2026) Application of Imaging Modalities in the Diagnosis of Branchial Anomalies. *Journal of Biosciences and Medicines*, **14**, 15-27.
https://doi.org/10.4236/jbm.2026.141002

Received: November 23, 2025
Accepted: December 28, 2025
Published: December 31, 2025

Abstract

BA (Branchial anomalies) are congenital malformations resulting from the incomplete regression of branchial apparatus structures during embryonic development. These lesions often exhibit complex anatomy and may adhere closely to critical vascular and neural structures in the neck. Given the rarity of these conditions and the consequent potential for limited clinical familiarity, selecting an appropriate imaging modality is crucial. Accurate identification of the internal opening, delineation of the fistula tract, and provision of reliable imaging evidence are essential to guide surgical planning, achieve complete excision of the pathological tissue, and prevent recurrence. This article reviews the current progress in various imaging techniques applied to the diagnosis of BA.

Keywords

Branchial Anomalies, Ultrasound, Computed Tomography (CT), Magnetic Resonance Imaging (MRI), Fiberoptic Laryngoscopy

1. Introduction

BA (Branchial anomalies) are a group of congenital malformations originating from the incomplete regression of the branchial apparatus during embryonic development [1] [2]. They represent the third most common congenital lesion in the pediatric head and neck region, following thyroglossal duct anomalies and lymph node pathologies [3] [4], accounting for approximately 30% of all congenital neck masses [5]. These lesions are predominantly distributed along the tract from the auricle to the clavicle, with a higher prevalence observed on the left side of the neck [6] [7].

Based on their embryonic origin and anatomical location, BA are primarily classified into four types. Second BA are the most common, accounting for approximately 90% of all cases, while third and fourth BA are relatively rare [3] [8]. Depending on the specific developmental defects of the branchial apparatus and the presence or absence of an opening between the neck and pharynx, they can be further categorized into branchial fistulas, sinuses, and cysts [2] [9]. A complete fistula refers to a tract with both internal (pharyngeal) and external (cervical) openings. If one or both ends are blind, it is termed a sinus or cyst, respectively [10]-[12]. These three forms are interconvertible. For instance, recurrent inflammation may lead to the formation of granulation tissue or scarring, which can obliterate one or both ends of a fistula, thereby converting it into a sinus or cyst [13] [14]. Radiologically, complete visualization of a branchial fistula is rare. Even if the tract is patent, the internal opening is often covered by a membranous structure, and it is uncommon for both openings to be simultaneously patent [11]. First branchial fistulas, also known as auriculocervical fistulas, are located above the hyoid bone, with their internal openings typically found in the external auditory canal, tragus, or mastoid region [3] [9]. Second branchial fistulas usually present with an external opening in the skin near the junction of the lower and middle thirds of the anterior border of the sternocleidomastoid muscle, while their internal opening is located in the tonsillar fossa [8] [15]. Third branchial fistulas course between the internal and external carotid arteries and open internally into the pyriform sinus, whereas fourth branchial fistulas are situated in the parapharyngeal space, closely adjacent to the pharyngeal wall, with their internal openings typically in the upper esophagus [4] [16]. Distinguishing between third and fourth BA can be challenging, partly due to ongoing controversies in their classification and their many overlapping features. Definitive differentiation often relies on surgical and histopathological findings [14]. Pyriform sinus fistulas include the traditional types of third and fourth branchial fistulas [17] [18]. Cysts of various types generally develop along the course of their corresponding fistulous tracts [19]. Accurate classification of BA primarily depends on the identification of the internal opening and the anatomical course. Therefore, imaging plays a crucial role in determining the type of anomaly, precisely locating the internal opening, differentiating between various subtypes, and guiding the formulation of an appropriate surgical strategy [20].

BA primarily presents with neck masses, recurrent infections, abscess formation, and drainage through sinuses [4] [21]. This condition predominantly affects children, with approximately 80% of cases diagnosed before the age of 5; however, the misdiagnosis rate is as high as 15% [8] [22] [23]. BA are uncommon and not well recognized by clinicians. Because they are often associated with infection, especially in the case of first, third, and fourth BA, they are often misdiagnosed as simple inflammatory diseases and managed only with incision and drainage. Studies indicate that about 73.3% of patients have undergone abscess incision and drainage, with an average diagnostic delay of 4 years [24]. Surgical excision is the gold standard for treating BA [25]. The key to surgery is the complete removal of the lesion, including all epithelial tissues such as the cyst, fistula tract, cyst wall, and both internal and external fistula openings, while carefully protecting adjacent critical blood vessels and nerves to prevent injury-related complications [13] [23]. For first BA, special attention must be paid to preserving the branches of the facial nerve; for third/fourth BA, it is essential to avoid injury to the recurrent laryngeal nerve and the thyroid gland [26]. Initial complete excision is crucial, as incomplete resection and inadequate management of the internal fistula opening are the main causes of postoperative recurrence and infection [1] [27]. However, approximately 50% of patients have previously undergone incomplete surgery [28]. The overall postoperative recurrence rate for BA ranges from 3% to 14%, while surgery performed during an active infection phase is associated with a significantly higher recurrence rate of 14% to 22% [8] [18] [23]. Diagnosis of BA is typically based on history (e.g., sinus drainage since birth), physical signs (e.g., neck mass, sinus tract), imaging studies, and fine-needle aspiration cytology (FNAC, which may reveal squamous epithelial cells or cholesterol crystals in cystic fluid) [4] [29]. The key to successful surgery lies in the complete excision along the entire fistula tract and secure ligation of the internal opening, making thorough preoperative imaging evaluation essential [11]. Currently, there is no universally recommended imaging technique for BA. Available options include ultrasonography, X-ray barium swallow fistulography, computed tomography (CT), magnetic resonance imaging (MRI), and fiberoptic endoscopy [6] [30] [31]. Therefore, this article aims to review the application of the aforementioned clinically used imaging techniques in the diagnosis of BA, systematically comparing and analyzing their respective advantages and limitations.

2. Role of Ultrasonography in the Diagnosis of BA

Ultrasonography can demonstrate fluid-filled tubular structures or gas shadows in the fistula region, along with inflammatory signs such as wall thickening and increased blood flow signals [18] [32]. Most scholars agree that preoperative B-mode ultrasound offers good diagnostic value for branchial cleft cysts, with a reported accuracy of up to 88.12% [12]. Conventional high-frequency ultrasound achieves a 100% detection rate for neck masses, with a sensitivity of 82.4% and specificity of 66.7% in diagnosing branchial cleft cysts. Owing to its advantages of being radiation-free, providing real-time imaging, and being easy to perform, it serves as a primary screening tool for BA [13] [33]. However, the diagnostic positivity rate of conventional ultrasound for BA is relatively low, with only about 10% of cases confirmed [24]. It is primarily used for initial screening during secondary acute infection to rapidly assess abscess extent and guide emergency interventions such as incision and drainage. During non-infectious phases, its diagnostic value for complex BA is very limited [34]. Furthermore, limited by its physical penetration depth, conventional ultrasound struggles to fully visualize deeply located or tortuous fistula tracts (e.g., long tracts extending to the pharynx, or third/fourth branchial fistulas), particularly in accurately locating the internal opening [6] [7]. The accuracy of ultrasonographic diagnosis is also highly operator-dependent, relying on the sonographer's familiarity with neck anatomy and knowledge of the various possible fistula variations.

Contrast-enhanced ultrasound (CEUS), typically performed via intravenous injection of contrast agents, allows precise assessment of tissue perfusion. Compared to CT contrast agents, ultrasound contrast agents have minimal side effects [35]. In recent years, intracavitary contrast-enhanced ultrasound (ICEUS) has been increasingly applied across various clinical specialties, accumulating substantial experience in diagnosing internal fistulas such as anal fistulas, biliary fistulas, and enteric fistulas associated with Crohn's disease [36]-[38]. Maximilian Rink *et al.* reported a case of a second branchial cleft fistula in which SonoVue contrast agent was injected preoperatively into the fistula tract via the external opening or subcutaneously. This enabled enhanced ultrasonographic visualization of the entire fistula tract and successfully demonstrated its connection to the tonsillar fossa (internal opening). The study highlighted that ICEUS provides crucial supplementary information for surgeons, such as the precise course of the fistula and its anatomical relationship to major neck vessels. This technique facilitates improved surgical planning, avoids radiation exposure, and eliminates the need for imaging under general anesthesia, offering particular benefits for pediatric patients [35]. However, intracavitary contrast-enhanced ultrasound (ICEUS) is an emerging technique, with limited clinical evidence and a scarcity of literature regarding its application in diagnosing branchial anomalies.

3. Application of Barium Swallow Radiography in the Diagnosis of BA

Barium swallow radiography allows real-time observation of contrast flow in the pharyngeal and cervical regions during barium ingestion, thereby enabling visualization of the entire fistula tract. Studies indicate that the fistula and its internal opening can be successfully demonstrated in approximately 50% - 64% of patients using esophageal barium radiography [13]. This technique achieves a diagnostic sensitivity of 80% for pyriform sinus fistula, with the typical imaging manifestation being a linear tract of contrast agent extending downward and forward from the medial wall of the pyriform sinus toward the thyroid region [18]. Peijun Zhang *et al.* suggest that for cases clinically highly suspicious of branchial fistula, barium swallow radiography may be regarded as the diagnostic gold standard, while CT and ultrasonography can serve as auxiliary localization tools [39]. However, Zhipeng Sun *et al.* hold a different view, stating that barium radiography can only confirm the presence of a fistula and is inadequate for evaluating its anatomical relationship with surrounding structures [26]. Additionally, barium swallow radiography involves ionizing radiation and must be used cautiously in pediatric or pregnant patients. It is also insensitive to branchial cysts or sinuses without an internal opening. Moreover, the examination should be performed during a non-infectious inflammatory period, as inflammatory exudates may obstruct the fistula tract during active infection, impeding adequate contrast filling.

4. Application of Computed Tomography in the Diagnosis of BA

CT examination can further delineate the complete fistula tract (from the cutaneous opening to the oropharyngeal lateral wall) and serves as an important supplementary diagnostic tool following conventional ultrasonography [6] [32]. First BA on CT typically appears as round hypodense shadows or tubular structures extending from the external auditory canal to the parotid gland or mandibular angle, often with wall enhancement. Second BA commonly presents as oval hypodense shadows posterior to the submandibular gland or tubular structures extending from the tonsillar fossa to the anterior border of the sternocleidomastoid muscle. Pyriform sinus fistula during quiescent phases may show punctate gas shadows or hypodense tracts at the upper thyroid pole, while during infection phases, CT reveals heterogeneous hypodense areas around the thyroid with wall enhancement [40]. Choi *et al.* propose that CT is a core tool for diagnosis and surgical planning [28]. In a retrospective study of 52 patients with second BA by Wei Chen *et al.*, CT demonstrated a diagnostic accuracy of 86.5% [34]. Further research by Zhou Yilong *et al.* indicated that CT achieved diagnostic accuracies of 87.4%, 84.2%, and 89.1% for first BA, second BA, and pyriform sinus fistula, respectively, establishing it as a gold standard for preoperative evaluation that significantly reduces misdiagnosis rates and surgical complications [40].

Contrast-enhanced CT involves the intravenous administration of iodinated contrast via arm injection following non-contrast scanning, with rapid arterial and venous phase imaging performed as the contrast circulates to cervical tissues. This technique achieves a diagnostic accuracy of 91.2% for pyriform sinus fistula [7] [41]. WANPENG LI *et al.* recommend enhanced CT as the preferred preoperative examination for first BA, as it clearly delineates the lesion extent and anatomical relationships with the facial nerve, external auditory canal, and parotid gland [24]. However, this technique primarily outlines the fistula tract or cyst wall through enhancement, which is susceptible to interference from surrounding inflammation, making it difficult to visualize the complete fistula course, fine branches, and internal opening.

Fistulography CT involves catheter-guided injection of iodinated contrast through the external skin opening of the fistula tract for direct visualization. On CT images, the fistula appears as a contrast-filled cord-like structure that is clearly distinguishable from adjacent neurovascular tissues, achieving a localization accuracy of 100% in diagnosing BA [6] [26]. This technique enables direct, three-dimensional, and complete visualization of the fistula's origin, course, branching pattern, and termination (internal opening), facilitating comprehensive preoperative assessment and avoiding unnecessary surgical exploration [42]. Multi-detector CT fistulography can further provide three-dimensional reconstructions [43]. J. Whetstone and Y. Bajaj *et al.* regard CT fistulography with iodinated oil as the gold standard for preoperative evaluation of first BA [23] [44], while Zhipeng Sun, Goff, and others also identify CT fistulography as the gold standard for defining fistula course and internal opening [26].

However, all CT techniques involve radiation exposure. Given that BA frequently occur in pediatric populations, radiation risk represents a significant clinical concern that cannot be overlooked [45].

5. Application of MRI in the Diagnosis of BA

A fistula resulting from BA is an abnormal soft tissue tract connecting the cutaneous surface (typically on the lateral neck) with the pharyngeal cavity or other deep structures [46]. Magnetic resonance imaging (MRI) offers significant advantages in the evaluation of such conditions, particularly due to its excellent soft tissue resolution. On T2-weighted or short-tau inversion recovery (STIR) sequences, the fistula typically appears as a linear hyperintense tract, presenting clear contrast with the surrounding tissues. In a case report of a 25-year-old female with a second branchial cleft fistula, Keogh *et al.* noted that preoperative MRI clearly demonstrated the fistula course and its anatomical relationship with adjacent neurovascular structures [22]. MRI can accurately assess the extent and pathway of BA, providing reliable evidence for surgical planning; hence, some scholars recommend it as a first-line imaging modality [30]. For preoperative diagnosis of pyriform sinus fistula, MRI achieves a positive predictive value of 92.3% [41]. When combined with gadolinium-based contrast for fistulography, it can clearly outline the entire fistula tract through abnormal enhancement, allowing precise localization of the internal opening. However, as pointed out by scholar Zhipeng Sun *et al.*, while MRI is sensitive for detecting branchial cysts, its performance in visualizing fistulous tracts is relatively inconsistent [26]. Moreover, the relatively long data acquisition time of MRI increases susceptibility to motion artifacts, and since BA frequently occurs in the pediatric population, the frequent need for sedation introduces associated risks. The high cost of MRI and limited availability of equipment also restrict its widespread clinical use to some extent [7]. Ahuja contends that MRI provides superior soft tissue contrast and is more advantageous than CT [47]. Some scholars argue that although MRI avoids the radiation exposure associated with CT imaging, it is seldom recommended as the preferred imaging method for evaluating BA, as CT scans are more readily accessible, faster, and generally better tolerated by young children [6].

6. Application of Fiberoptic Laryngoscopy in the Diagnosis of BA

In otorhinolaryngology practice, electronic fiberoptic laryngoscopy is commonly used to examine the oropharynx and hypopharynx in search of internal fistulous openings associated with BA. For patients in whom the internal fistula opening is not identified preoperatively, intraoperative exploration with supporting laryngoscopy is recommended to improve the detection rate [33]. During the procedure, the laryngoscope can be gently advanced from the external fistula opening, and methylene blue or other contrast agents may be injected through the laryngoscope channel to aid visualization. The Valsalva maneuver can also assist in identifying the internal fistula opening. Studies have shown that laryngoscopy performed during the quiescent inflammatory phase has a positive predictive value of 75.6% for pyriform sinus fistula [41]. In a retrospective study involving 10 patients with fourth branchial cleft anomalies, Wan-Xin Li *et al.* recommended the combined use of preoperative laryngoscopy and barium swallow radiography to accurately localize the internal opening, assess the fistula course, and visually examine the fistula pathway, internal structure, and internal opening position, thereby enabling precise ligation of the internal orifice under laryngoscopic guidance [45] [48]. However, laryngoscopy requires general anesthesia and is considered a minimally invasive procedure. It is difficult to insert the scope into completely obliterated or highly stenotic fistulas, and the limited field of view restricts comprehensive visualization of the entire fistula tract and its anatomical relationship with cervical vessels and nerves [33].

7. Application of Multimodal Image Fusion Technology in the Diagnosis of BA

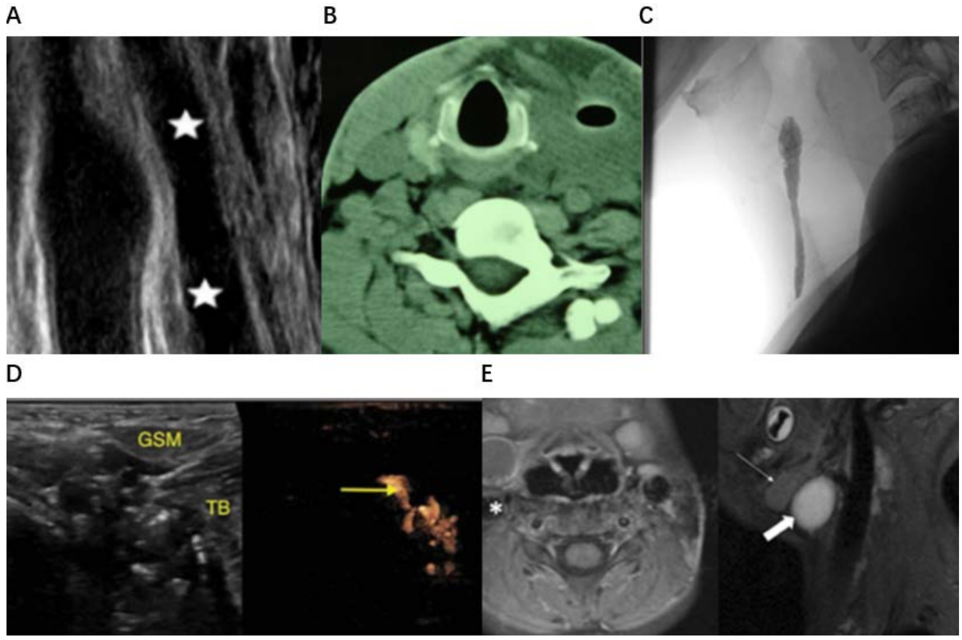
The combination of multi-slice spiral computed tomography (MSCT) and multiplanar reformation (MPR) enables clear visualization of the location of branchial cysts, the course of fistulous tracts, and their anatomical relationships with critical structures such as the carotid sheath, thyroid gland, and facial nerve, thereby effectively preventing intraoperative injury. With a qualitative accuracy of 99.9% and a localization accuracy of 100.0%, this technique is regarded as the preferred imaging modality for the diagnosis and preoperative evaluation of BA in children [49]. Its core advantage lies in the ability to provide three-dimensional reconstructions that intuitively display the stereoscopic anatomy of the lesions, offering surgeons precise images of fistula courses and their surrounding relationships, which holds significant guiding value for clinical diagnosis and surgical planning, though related literature reports remain relatively limited at present. Multimodal image fusion technology demonstrates significant potential in diagnosing branchial anomalies by integrating the advantages of CT, MRI, and ultrasonography. Its clinical value lies in the three-dimensional visualization of complex fistula pathways and precise localization of adjacent neurovascular structures, thereby optimizing surgical planning and reducing the risk of iatrogenic injury. However, the technology faces challenges such as registration errors caused by soft tissue deformation in the neck, high hardware costs, and a steep learning curve. Future efforts should focus on developing more intelligent registration algorithms and promoting clinical translation research.

8. Summary and Prospects

Comprehensive evaluation of branchial cleft malformations typically requires the integration of multiple imaging modalities, including ultrasonography (USG), computed tomography (CT), and magnetic resonance imaging (MRI). Fistulas of BA typically appear as tubular anechoic or mixed-echoic structures on conventional ultrasound, tubular hypodense tracts on CT images, and exhibit hypointensity on T1-weighted images and hyperintensity on T2-weighted images on MRI. Enhanced imaging may reveal wall enhancement and surrounding inflammatory reactions, while fistulography demonstrates contrast-filled tubular hyperechoic or hyperdense tracts (**Figure 1**). Ultrasonography serves as a first-line screening tool, offering advantages such as absence of radiation and operational convenience, making it particularly suitable for the initial assessment of benign cystic lesions in children. CT and MRI, on the other hand, provide superior anatomical detail, clearly delineating the fistula tract and its complex relationships with surrounding structures such as the parotid gland, facial nerve, major cervical vessels, and laryngeal nerves. These modalities are essential for preoperative classification, localization, and surgical planning. In identifying the internal fistula opening, barium swallow radiography and CT fistulography effectively demonstrate its connection to pharyngeal structures like the pyriform sinus. Contrast-enhanced fistula sonography, as a safe and radiation-free technique, is not only convenient but also capable of accurately outlining the fistula course and locating the internal opening. Although its current clinical application is limited, it holds promise for playing a more significant role in the diagnosis of BA in the future. Intraoperative endoscopic visualization provides definitive confirmation of the internal opening (**Table 1**). Histopathological examination—characterized by a cyst lining of stratified squamous or ciliated columnar epithelium, surrounding lymphoid tissue infiltration, and inflammatory cell exudation in cases of infection or rupture—remains the gold standard for definitive diagnosis [4] [29].

Table 1. Comparison of imaging modalities for diagnosing branchial anomalies.

Imaging Modality	Advantages	Disadvantages
Ultrasound	No radiation, real-time imaging, portable Suitable for initial screening High accuracy for branchial cysts (88.12%) ICEUS enhances fistula visualization without radiation	Poor visualization of deep or tortuous fistulas It is difficult to locate internal openings Low diagnostic yield (only 10%) Operator-dependent
Barium Swallow	Real-time visualization of the fistula tract and internal opening High sensitivity for pyriform sinus fistula (80%) Considered the gold standard by some authors	Ionizing radiation Cannot assess the surrounding anatomy Insensitive for cysts/sinuses without internal opening Must be performed in the non-inflammatory phase
CT	Clear depiction of the entire fistula and adjacent structures High diagnostic accuracy (86.5% - 91.2%) 3D reconstruction with fistulography; nearly 100% localization accuracy Recommended for surgical planning.	Ionizing radiation, caution in children Limited to small branches and internal openings Susceptible to inflammatory artifacts
MRI	No radiation, excellent soft tissue contrast Clearly shows a relationship with neurovascular structures High PPV for pyriform sinus fistula (92.3%) Ideal for complex cases.	Long scan times, motion artifacts Often requires sedation in children High cost, limited availability Inconsistent visualization of fistulas
Fiberoptic Laryngoscopy	Direct visualization of the internal opening Can be combined with dye or the Valsalva maneuver Intraoperative assistance for locating the internal opening	Requires general anesthesia, invasive Cannot show the entire fistula or surrounding structures Difficulty in obliterated or narrow fistulas



(A) Coronal ultrasound view of the upper neck demonstrates the fistulous tract (asterisk) [7]. (B) Left-sided infected fourth branchial fistula, with the tract visible at the center of the inflammatory mass [10]. (C) Sagittal CT fistulogram shows contrast enhancement within the anterior cervical sinus tract, ending blindly at the C5 - C6 vertebral level [31]. (D) Contrast-enhanced ultrasound fistulography clearly delineates the course of the fistula tract [35]. (E) Axial fat-suppressed T1-weighted postcontrast and sagittal short tau inversion recovery (STIR) MR images reveal a round, well-defined lesion that is isointense on T1-weighted images, hyperintense on T2-weighted images, and shows thin peripheral enhancement (thick white arrow). The lesion is located anterior to the sternocleidomastoid muscle and carotid sheath (asterisk) [5].

Figure 1. Imaging manifestations of branchial anomalies.

With continuous advancements in medical imaging technology, the combined application of ultrasonography, CT, and MRI has significantly compensated for the limitations of single imaging modalities, thereby improving diagnostic accuracy. For patients with BA, ultrasonography is the preferred initial modality for screening and differential diagnosis. CT or MRI can then be used to further clarify the fistula tract. Preoperative assessment relies on CT fistulography or contrast-enhanced ultrasonography via the fistula to visualize the entire course and identify the internal opening. Intraoperatively, endoscopy provides real-time guidance to avoid critical neurovascular structures, ensuring complete lesion excision and secure ligation of the internal opening. Optimizing the strategy for combined imaging will further enhance preoperative diagnostic efficacy, establish a more solid foundation for developing individualized treatment plans, effectively reduce the risk of surgical complications, avoid overtreatment and disease recurrence, and ultimately advance the diagnosis and treatment of branchial anomalies toward greater precision and minimally invasive approaches.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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