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Ultrasound characteristics of alveolar soft part sarcoma in pediatric patients: a retrospective analysis

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Abstract

Objective This study aims to review and summarize the ultrasound characteristics of alveolar soft part sarcoma (ASPS) in children.

Methods We retrospectively analyzed 20 pediatric ASPS cases confirmed by surgery or biopsy at our hospital between January 2014 and January 2024. Clinical data, including age, sex, symptoms, and tumor location, were collected. Ultrasound reports and images were reviewed to extract data on tumor size, boundaries, echogenicity, and vascularity.

Results The study included 20 children with ASPS. The tumors were located in the trunk and limbs (50%), as well as in the head and neck (50%). Compared with tumors in the trunk and limbs, head and neck tumors were smaller in size, had more pronounced symptoms, and had a lower incidence of metastasis. Ultrasound features predominantly included hypoechoic masses with clear boundaries, heterogeneous echogenicity, and rich internal and surrounding vascularity, often with tortuous and dilated blood vessels. Eight patients had distant metastases at diagnosis, seven of which involved the lungs. There was a moderate correlation between tumor size and the risk of distant metastasis ($r = 0.64$).

Conclusion Understanding the clinical and ultrasound characteristics of pediatric ASPS can facilitate earlier and more accurate diagnosis.

Keywords Alveolar soft part sarcoma (ASPS), Pediatric, Ultrasound, Diagnosis

Background

Soft tissue sarcomas (STSs) are rare malignant tumors, with the most common types being undifferentiated pleomorphic sarcoma, liposarcoma, and leiomyosarcoma [1]. Alveolar soft part sarcoma (ASPS) is particularly rare, accounting for only 0.2–0.9% of all STS cases [2]. Most of the literature on ASPS consists of case reports and series, indicating its unknown tissue origin, aggressive nature, tendency to metastasize, and poor prognosis [3].The 5-year disease-specific survival was 68%, and metastasis was the only adverse prognostic factor [4]. Commonly affected sites include the extremities, bladder, breast, larynx, cervix, and bones. However, in children, there is a significant increase in the incidence of ASPS in the head

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and neck region [5]. Most of our knowledge about ASPS is based on studies in adults [6], as pediatric studies are limited due to its rarity. Studies suggest that pediatric ASPS may exhibit less aggressive behavior and distinct biological characteristics, resulting in a better prognosis than in adults [7].

Recognizing the imaging characteristics of ASPS is crucial for the diagnosis, staging, and early treatment of this slow-growing tumor [8]. Current imaging studies on ASPS focus mainly on magnetic resonance imaging (MRI) and computed tomography (CT) [9]. There are few studies on ultrasound, which are mostly limited to case reports [10, 11]. This study aims to analyze the clinical and ultrasound characteristics of pediatric ASPS before treatment to aid in accurate preoperative diagnosis and determine a subsequent treatment plan.

Materials and methods

Study population

We retrospectively reviewed 20 pediatric ASPS cases confirmed by pathology at our hospital from January 2014 to January 2024. All patients underwent ultrasound examination of the lesion before treatment, and their clinical data were complete. This study was approved by the institutional ethics review committee of Beijing Children's Hospital.

The exclusion criteria were as follows: (a) no ultrasound examination before consultation. (b) treatment received at other hospitals before consultation.

The clinical data collected included age, sex, symptoms, tumor location, and treatment details.

Ultrasound examination

Ultrasound examinations were performed via a Hitachi Ascendus ultrasound system with C715 (1–5 MHz) and L52 (3–7 MHz) probes. Two experienced ultrasound physicians (with more than ten years of experience) reviewed all the ultrasound images and short loops. In case of any discrepancies, the final opinion will be provided by the department's chief physician.

The selection of key features is informed by our clinical experience as well as relevant literature [12], included the following: (a) Tumor size: The maximum diameter was measured on the basis of previous reports and images. (b) Boundaries: Clear boundaries if the tumor edges are well defined. (c) Echogenicity: Compared with the surrounding muscle, the echogenicity is defined as hypoechoic, isoechoic or hyperechoic. (d) Echo characteristics: Homogeneous if internal echoes are uniform; otherwise, heterogeneous. (e) Vascularity: Less than 1/2 of the area with blood flow signals is not rich, and more than 1/2 of the area is rich. (f) Arteriovenous fistulas: Spectral Doppler may show reduced resistance and increased flow velocity in the inlet region of the fistula.

Statistical analysis

The data distribution characteristics were initially summarized via Q–Q plots. Normally distributed variables are expressed as the means ± standard deviations, and non-normally distributed variables are expressed as medians (interquartile ranges). Categorical data are expressed as counts (percentages). Spearman correlation analysis was used to assess the relationship between the ultrasound characteristics of the tumor and the risk of metastasis, with correlation coefficients indicating the risk of distant metastasis.

Results

All 20 ASPS patients were pathologically confirmed, and their demographic data are summarized in Table 1. Fourteen patients were female, and six were male, with a median age of 7 years (range 2–15 years). Seven patients (35%) had clinical symptoms, including snoring (3 patients), unclear speech (1 patient), vomiting (1 patient), and bleeding (2 patients). There were also 4 cases of tumors in the limbs, which parents mostly described as painless, slowly growing masses.

The average tumor size was 5.84 cm (range 1.5–20.2 cm). There were 10 cases (50%) of tumor lesions located in the trunk and limbs, including 1 case (5%) in the psoas major muscle, 3 cases (15%) in the retroperitoneum, 3 cases (15%) in the upper limbs, 1 case (5%) in the left lower limbs, 1 case (5%) in the bladder, and 1 case (5%) in the vagina. The tumor lesions were located in the head and neck in 10 patients (55%), including 2 patients (10%) in the Neck Muscles, 2 patients (10%) in the pharynx, and 6 patients (30%) in the tongue. Compared with trunk and limb tumors, head and neck tumors occur in younger children, are smaller, have more pronounced symptoms, and have a lower incidence of metastasis (Table 2).

The ultrasound features (Figs. 1, 2 and 3) included Hypoechoicity (18/20), heterogeneous echoes (16/20), clear boundaries (18/20), and rich internal vascularity (20/20), with tortuous and dilated vessels observed inside and around the tumor (20/20). Larger tumors correlated with less defined boundaries ($r=0.52$) and more heterogeneous internal echoes ($r=0.57$) (Fig. 4).

We reviewed the pathological reports of all the patients, and 7 patients (35%) presented hemorrhagic necrosis, including 1 patient with a range greater than 80% calcification. Ultrasound also revealed liquefaction and calcification, while the remaining 6 patients showed no significant changes on ultrasound. Three patients

Table 1 Summary of Clinical Information for all patients

Patient *n°*	Age	Sex	Location (Side)	Symptom	Longest Diameter(cm)	Metastasis* (Sites)	Treatment
1	6	F	Lumbar Muscle (R)	No	6.5	Yes (Lung)	Excision
2	11	F	Retroperitoneal (L)	No	20.2	Yes (Lung Vertebra)	Excision
3	13	M	Retroperitoneal (L)	No	17.2	Yes (Lung)	Excision
4	8	F	Retroperitoneal (R)	No	6.9	Yes (Lung Lymph node)	Excision Targeted drugs
5	5	F	Forearm muscle (R)	No	3.5	No	Tumor embolism Excision
6	14	M	Forearm muscle (R)	No	8.5	Yes (lung)	Excision
7	15	F	Thigh muscles (L)	No	11.7	Yes (lung)	Tumor embolism Excision
8	13	M	Upper arm muscles (L)	No	3.1	No	Excision
9	12	F	Trigone of bladder	No	2.4	No	Excision
10	8	F	Vagina	Vaginal Bleeding	3.1	Yes (Lymph node)	Uterine artery embolism
11	3	F	Parapharyngeal (R)	Snore	3.6	No	Excision
12	4	F	Oropharynx	Snore	2.6	Yes (Lung)	Excision
13	6	F	Neck Muscles (Back)	No	3.2	No	Excision
14	4	F	Neck Muscles (L)	No	1.5	No	Excision
15	7	M	Tongue	Pronunciation difficulties	4.5	No	Excision
16	7	F	Tongue	Vomit	2.6	No	Excision
17	2	M	Tongue	Bleeding	2.8	No	Tumor embolism Excision
18	3	M	Tongue	Snore	5.4	No	Excision Radiotherapy
19	9	F	Tongue	No	3.0	No	Excision
20	4	F	Tongue	No	4.5	No	Tumor embolism Radiotherapy

*Representing lung metastasis, vertebral metastasis, and lymph node metastasis

Table 2 Summary of the clinical and ultrasound characteristics of tumor lesions in different locations

			Head Neck	%	Trunk Limb	%	*P*
Clinical	Number	Median	10	50	10	50	0.001
Ultrasonic	Age	F	5	70	10	70	1.000
	Sex	M	7	60	7	10	0.018
	Symptom	Y	3	10	3	70	0.018
	Metastasis	N	6	100	1	80	0.024
	Longest Diameter(cm)	Y	4	90	9	90	0.151
	Boundary	N	1	70	7	90	1.000
	Echo	Clear	9	100	3	100	0.288
	CDFI	Unclear	3.37 ± 1.15	100	8.31 ± 6.23	100	1.000
		Hypoechoic	10		8		1.000
		Isoechoic	0		2		
		Heterogeneity	9		9		
		Homogeneity	1		1		
		Hypervascularity	7		9		
		Rich blood perfusion	3		1		
			10		10		
			10		10		

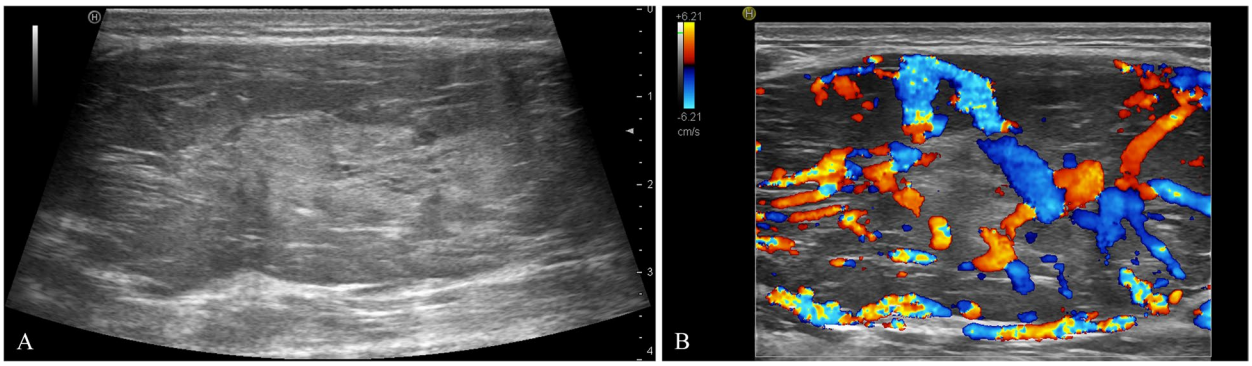

Fig. 1 **A** Gray-scale ultrasound revealed a hypoechoic mass within the muscle layer of the left upper limb, with clear boundaries and heterogeneous internal echoes (**B**) Color Doppler imaging showing multiple tortuous blood vessels inside the mass, with rich blood flow signals

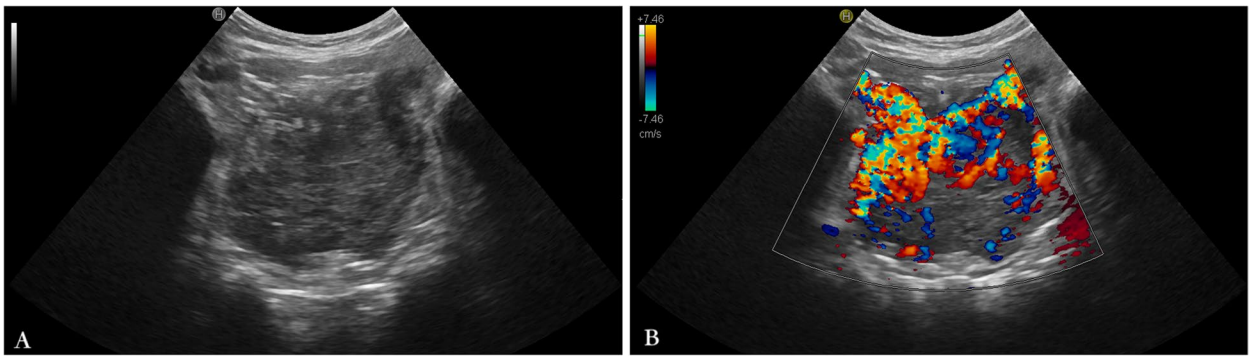

Fig. 2 **A** Gray-scale ultrasound revealed a hypoechoic mass within the soft tissue of the tongue root, with clear boundaries and heterogeneous internal echoes (**B**) Color Doppler imaging showing multiple tortuous blood vessels inside and around the mass, with rich blood flow signals

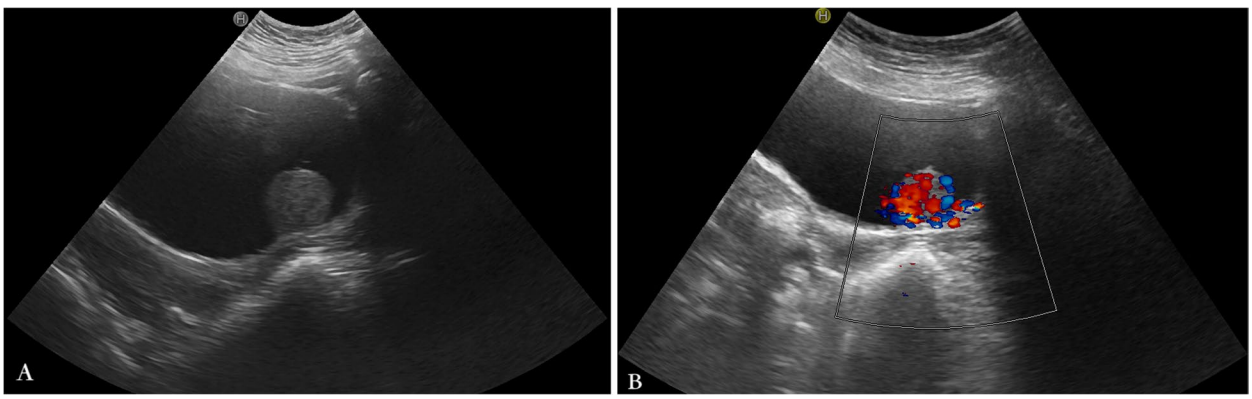

Fig. 3 **A** Gray-scale ultrasound revealed a hypoechoic mass with clear boundaries and heterogeneous internal echoes in the bladder. (L) (**B**) Color Doppler image showing rich blood flow signals inside the mass, extending into the bladder wall. (R)

presented with tumor thrombi, but ultrasound revealed no relevant changes.

Among the 20 patients, 8 (40%) had distant metastases at the time of diagnosis. Specifically, 7 patients had pulmonary metastases, and 3 patients had metastases to regional lymph nodes or vertebral bodies. Among those with metastases, 6 patients (6/8) had a primary tumor diameter greater than 5 cm. There was a moderate

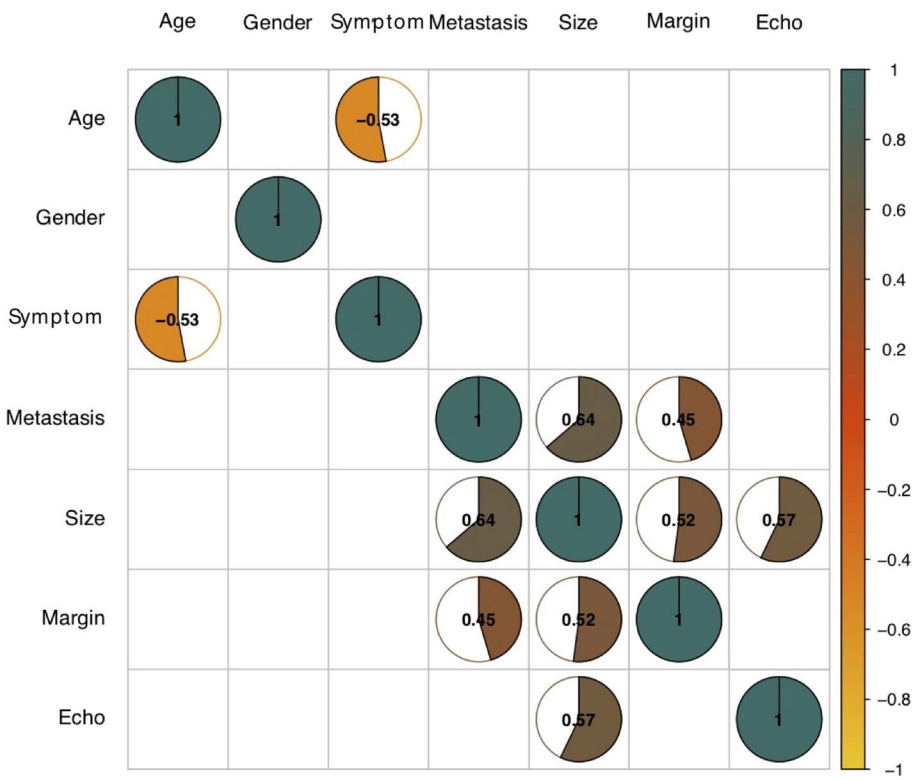

Fig. 4 The horizontal and vertical coordinates contain epidemiological characteristics and ultrasound observation indicators; The horizontal and vertical intersection points indicate the strength of the correlation (Spearman correlation coefficient), whereas the blank area indicates that there is no significant statistical significance in the correlation

correlation ($r=0.64$) between the maximum diameter of the primary tumor at diagnosis and the occurrence of distant metastases. A larger tumor size indicated a greater risk of metastasis (Fig. 4).

In this group of patients, 15 children (75%) underwent direct surgical resection, while 5 children (25%) received sclerotherapy at the initial discovery of the tumor, of which 3 children opted for surgical resection after observation. As of the submission date of the manuscript, there are still two children who have not undergone surgery. Both are currently under regular ultrasound monitoring (one case shows no change in tumor size, while the other shows a slight increase in tumor size).

Discussion

ASPS is extremely rare in children, comprising 1–2% of all pediatric soft tissue sarcomas [13]. It typically progresses slowly and has a high metastatic rate. Compared with adults, children with ASPS exhibit distinct clinical characteristics. The most significant difference lies in the primary site of onset: in adults, ASPS originating from the head and neck region accounts for only 3.4%, whereas in children, this proportion can reach 32% [14]. Among younger children, ASPS predominantly affects the head and neck, whereas in older children, it tends to occur in the trunk and limbs. In our group of ASPS cases, 50% were located in the head and neck area. These findings further underscore that pediatric ASPS patients disproportionately involve the head and neck compared with adults.

Previous studies have shown a female-to-male ratio of 2:1 in the general population, but gender differences in children are less pronounced [15]. In our case series, the female-to-male ratio was 14:6, indicating a higher proportion of females, although more cases are needed for further validation. ASPS typically presents as a slow-growing, painless soft tissue mass that rarely causes functional impairment, leading to delayed detection [16]. Similarly, the older children in our group presented these characteristics. However, younger children, owing to the prevalence of head and neck lesions, tend to present earlier with related symptoms [17]. In our series, 6 patients (75%) with lesions in the oropharyngeal region presented symptoms, primarily related to compression.

The ASPS metastasis rate is high, with studies reporting a rate of 59% in adults [18], whereas in this group of children, the overall rate is 40%, which is lower than that in adults. With respect to the locations of metastasis,

in adults, metastases are most commonly found in the lungs, brain, bones, liver, and lymph nodes. In this group of children, the primary site of metastasis was the lungs (7/8), with 2 patients showing metastasis to surrounding lymph nodes and one child developing vertebral metastasis two years after onset. There is also a significant disparity in the metastasis rates depending on the location of the primary tumor. Compared with tumors in the trunk and limbs (70%), tumors in the head and neck have a low metastasis rate (10%). Previous reports have also indicated that the rates of ASPS metastasis from head and neck tumors (40%) are lower than those from tumors in the limbs (73%). The reasons for this may be related to the specific characteristics of head and neck lesions, where symptoms appear earlier and more prominently, leading to earlier medical intervention. In this group of patients, there was a significant correlation between tumor size and the metastasis rate, with larger tumors indicating a greater risk of metastasis. This finding aligns with previous research conclusions [19].

Ultrasound is often the initial examination in clinical practice for assessing soft tissue masses. It allows for rapid acquisition of information regarding the internal composition of the mass and accurately identifies the specific layer within the soft tissue tumor (such as fat, muscle, or fascia). This information is crucial in guiding subsequent clinical decisions, including regular follow-up, CT, MRI, or biopsy. Although ASPS is rare and difficult to diagnose, it has distinctive ultrasound characteristics. Despite significant clinical differences between head and neck lesions and those in the trunk and limbs, the ultrasound features of tumors in different locations are generally consistent. The most common ultrasound presentation is a well-defined hypoechoic lesion, which is consistent with previous ultrasound studies [8]. Lesions often display heterogeneous internal echogenicity; larger tumors may present areas of hemorrhage and necrosis, whereas smaller tumors tend to be relatively homogeneous. This group exhibited lesions with homogeneous echogenicity, with an average maximum diameter of only 2.4 cm. Most lesions had clear boundaries, although unclear margins were observed only in exceptionally large tumors. Tumors in limb muscles are typically spindle shaped, whereas those in the oral cavity tend to be round, possibly reflecting differences in tumor texture and surrounding tissue compression.

In comparison to the gray-scale ultrasound findings of ASPS, the color Doppler ultrasound characteristics are notably more distinctive. ASPS tumors typically exhibit rich intralesional and perilesional vascularization, characterized by tortuous and dilated vessels. In our series, varying degrees of tortuous and dilated vessels were observed both within and around all 20 lesions, with vessel diameters ranging from 0.1 to 2 cm. Some intralesional vessels show signs of proliferation and fusion, which is consistent with imaging findings [20]. However, due to the limitations of retrospective studies, the spectral information in our series was incomplete. In this group of cases, three children exhibited ultrasound findings of arteriovenous fistula. Color Doppler ultrasound showed bright and colorful blood flow signals in the fistula region during systole, while Spectral Doppler demonstrated decreased resistance and increased flow velocity at the fistula inlet, with arterialization of the venous spectral signals at the outlet. Additionally, one child underwent contrast-enhanced ultrasound, which revealed uniform moderate to high enhancement with a “fast in, slow out” pattern. Related studies have also reported similar results of ASPS in angiography [21]

ASPS is characterized by multiple malformed blood vessels and extremely rich blood flow signals, which can help differentiate it from lipomas and most soft tissue sarcomas [22]. However, ASPS is highly likely to be misdiagnosed as a vascular tumor or vascular malformation in deep tissues. Among our cohort of 20 pediatric cases, ultrasound provided diagnostic inclinations in 13 cases preoperatively, with 9 cases misdiagnosed as vascular tumors or malformations, highlighting their similarity. Both ASPSs and vascular tumors or malformations can present with tortuous dilated vessels internally and peripherally. Compared with the rare occurrence of ASPS, diagnosing vascular tumors or malformations is evidently easier. Ultrasound features aiding in differentiation include partly unclear boundaries [23], partially increased internal echoes (related to higher intratumoral fat content), and a significantly greater incidence of venous phleboliths than does the ASPS [24]. Peripheral and distant metastasis may indicate a greater possibility of ASPS, particularly pulmonary metastases (7/8), which strongly suggest the disease when it is identified alongside highly vascular soft tissue lesions on examination. Based on the characteristics of our own cases and previous literature, we selected relatively common pediatric soft tissue tumors that need to be differentiated from ASPS in clinical practice, and we organized and compared the main ultrasound features of each tumor (Table 3).

Due to the rarity of ASPS, most cases are often advised to undergo regular follow-up examinations when first discovered [25]. In cases where there is rapid growth or significant changes in the condition, it is recommended that the child undergo further imaging studies, including MRI of the local soft tissues and CT of the lungs, to assist in diagnosis. When there is a high suspicion of ASPS, especially in the presence of metastasis, we still prefer to obtain a definitive

Table 3 Ultrasound characteristics of ASPS and other common soft tissue tumors in children

	Gray Scale				Color Doppler		
	Echo		Boundary	VS	AVF	VM	BF
ASPS	Hypoechoic	Heterogeneity	Clear	No	Yes	Yes	Extremely rich
BVL [20, 32]	Hyperechoic	Heterogeneity	Unclear	Yes	Yes	Yes	Extremely rich
Lipoma [33, 34]	Isoechoic	Homogeneity	——	No	No	No	Poor
DT [35, 36]	Hypoechoic	——	Unclear	No	No	No	Rich
RMS [37, 38]	Hypoechoic	Heterogeneity	Clear	No	No	No	Rich

ASPS Alveolar soft part sarcoma, *BVL* Benign vascular lesions, *DT* Desmoid tumors, *RMS* Rhabdomyosarcoma, *VS* Venous stone, *AVF* Arteriovenous fistula, *VM* Vascular malformation, *BF* Blood flow

The blank space indicates that the manifestations of this disease are diverse and lack distinct characteristics

diagnosis through biopsy as soon as possible, allowing the child to be quickly referred to our hospital's oncology center for treatment. Of course, prior to the biopsy, we conduct a thorough assessment, which includes: (a) detailed communication with the child's parents, explaining the risks and obtaining written consent; (b) ensuring the child's coagulation function is completely normal; (c) using the thinner possible needle (usually 18-20G) during the procedure and obtaining 2–3 tissue strips based on the integrity of the tissue sample. In this group, 8 cases with lesions in the limbs or retroperitoneum underwent ultrasound-guided percutaneous biopsy, and no significant bleeding or other complications were observed postoperatively. Other studies suggest that fine-needle aspiration can also provide sufficient evidence for disease classification, with potentially less damage. However, we do not have such experience, and further validation is needed [26]. .

Children with localized ASPS generally have a favorable long-term prognosis, with an overall 5-year survival rate of approximately 90% [27]. The current preferred treatment for ASPS involves wide surgical excision aiming for tumor-free margins, typically requiring 1–1.5 cm of tumor-free tissue surrounding the tumor bed [28]. For areas that are difficult to excise completely, such as the base of the tongue or adjacent to the pharynx, postoperative adjuvant therapy may be considered to prevent recurrence and metastasis [29]. Currently, targeted therapy shows great promise for children with unresectable or multiple metastatic disease. Many studies indicate that the prognosis of pediatric ASPS is better than that of adults, and the survival rate is higher in younger patients [30]. In our group, there is only one known case of death. Additionally, three pediatric patients in this group underwent surgical resection after tumor embolization. This method helps to reduce intraoperative bleeding from the tumor, which has been applied in surgeries involving other hypervascular tumors [31].

This study also has several limitations. Firstly, due to the rarity of the disease, the number of patients included is small, and the slow progression of the disease over a long period makes it difficult to track some cases, resulting in challenges in collecting prognosis and survival data. Secondly, the strong capabilities of the head and neck surgery department at our hospital may attract a higher number of pediatric patients with head and neck tumors, leading to selection bias. Furthermore, this study is a retrospective analysis that relies on archived ultrasound images and videos, which limits the acquisition of detailed imaging information. Additionally, the settings for color Doppler parameters cannot be standardized uniformly across cases. Despite these limitations, within the context of ASPS being an ultrarare sarcoma, our study represents one of the largest pediatric ultrasound-related research efforts to date.

Abbreviations

ASPS	Alveolar soft part sarcoma
STSs	Soft tissue sarcomas
MRI	Magnetic resonance imaging
CT	Computed tomography
CDFI	Color doppler flow imaging
BVL	Benign vascular lesions
DT	Desmoid tumors
RMS	Rhabdomyosarcoma
VS	Venous stone
AVF	Arteriovenous fistula
VM	Vascular malformation
BF	Blood flow

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Authors' contributions

S.W. wrote the main manuscript text, Q.R. and Y.H. were in charge of collecting clinical data, J.X. verified the pathology results, Y.W. conducted statistical analysis, L.J. and X.W. reviewed and modified the manuscript.All authors reviewed the manuscript.

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Data availability

"The datasets used or analyzed during the current study are available from the corresponding author upon reasonable request. Most of the data is provided within the manuscript.

Declarations

Ethics approval and consent to participate

All methods were carried out in accordance with the Declaration of Helsinki and the relevant guidelines/regulations. This study was approved by the institutional ethics review committee of Beijing Children's Hospital [2024]-E-112-R. Informed consent was waived by our Institutional Review Board because of the retrospective nature of our study.

Consent for publication

Since the database is de-identified, the requirement for consent to publish personal information about an individual was waived.

Competing interests

The authors declare no competing interests.

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