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Case Study

Histologically confirmed pediatric extracardiac rhabdomyoma: case series

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Abstract
Rhabdomyoma is a rare benign tumor of striated muscle, which can be either cardiac or extracardiac. Extracardiac rhabdomyomas can occur throughout the body, though the fetal and adult subtypes are most commonly found in the head and neck region.
We present three pediatric cases of extracardiac rhabdomyoma, fetal subtype, detailing their clinical presentations, computed tomography imaging, and tissue biopsy findings. Given the very rare occurrence of extracardiac rhabdomyoma and its relatively benign nature, histological diagnosis is crucial. In all three cases reported here, a diagnosis of extracardiac rhabdomyoma was confirmed, and treatment with local excision resulted in favorable outcomes.

Keywords Rhabdomyoma · Retroperitoneum · Surgery · Pediatrics

1 Background

Rhabdomyoma is a rare benign soft tissue tumor of skeletal muscle differentiation, accounting for only 2% of skeletal muscle tumors [1]. It can be classified as either cardiac or extracardiac rhabdomyoma [2, 3].

Extracardiac rhabdomyomas are further divided into three subtypes: adult rhabdomyomas, fetal rhabdomyomas, and genital rhabdomyomas. The fetal subtype exracardiac rhabdomyoma typically arises in striated muscle, most often in children under 3 years of age, with a predilection for the head and neck region [4]. Histological confirmation is crucial to differentiate rhabdomyoma from malignant soft tissue and skeletal muscle tumors. Rhabdomyoma is usually treated with local surgical excision, and recurrence is rare following complete removal [5, 6].

Pre-operative diagnosis based solely on clinical presentation and imaging is challenging. In this report, we present three cases of rhabdomyoma originating from the pharynx, retroperitoneum, and post-auricular region.

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2 Case presentation

2.1 Case one

A 3-year-old female thriving child presented to our tertiary hospital with right lateral neck swelling of five- months duration. Physical examination revealed an acutely sick-looking child in respiratory distress with a pulse rate of 120 beats/minute, respiratory rate of 28 breaths/minute, and maintained saturation of oxygen with atmospheric air. There was a 6 by 4 cm soft, non-tender, immobile mass over the right anterolateral neck with a smooth surface and regular border. She had grade II tonsillar enlargement and audible stridor, but no lymphadenopathy. Complete blood count and chest X-ray were normal. Neck ultrasound showed a right-side solitary enlarged mid-anterior cervical lymph node measuring 4.04 × 3.01 × 3.91 cm with heterogeneous density. A post-contrast head and neck computed tomography (CT) showed a homogenously enhancing defined right-sided mass lesion centered in the retropharyngeal space extending superiorly into the nasopharynx blunting the right fossa of Rosenmüller and inferiorly to the hypopharynx with narrowing of the right pyriform sinus. The mass measures 6 * 3.8 * 3.7 cm in size and has significantly narrowed the oropharynx with mass effect over the right carotid space displacing it posterolaterally (Fig. 1A–C).

She was kept with the diagnosis of partial upper airway obstruction secondary to retropharyngeal mass, and transcervical tumor excision was done. The biopsy showed a well-circumscribed lesion with no invasive growth pattern comprising bland oval to spindle cells having eosinophilic cytoplasm with cross striation arranged in fascicles with frequent rhabdomyoblastic differentiation with no myxoid stroma and necrosis. There were no mitosis and immature mesenchymal components. The histologic findings were consistent with the fetal (cellular) type of rhabdomyoma (Fig. 2A–C). The child is doing well on her second-year post-surgery follow-up after the complete surgical excision.

With these histologic findings a diagnosis of fetal rhabdomyoma, intermediate type was made.

2.2 Case two

A 2-year-old and 9 month-old female child presented to our hospital with painless swelling over the back of her right ear since the first week of life with rapid growth over one month with offensive discharge. Physical examination showed a huge pedunculated, fungating firm mass over the right posterior auricle with pus and blood mixed discharge. A head and neck CT scan showed a large heterogeneously enhancing soft tissue mass lesion centered in the right retro auricular [mastoid] region measuring 7.5 cm*6.0 cm*6.3 cm (AP*TR*CC), invading and pushing the pinna anteriorly (Fig. 3A, B). The mass had no evidence of bone erosion or intracranial extension. There were also multiple small well-defined enhancing soft tissue nodules in the right parietal and occipital scalp, the largest measuring 8 mm.

The patient underwent complete surgical excision and the biopsy showed well-circumscribed, not encapsulated, sheets of large cells, round to polygonal, with abundant eosinophilic fibrillar or granular cytoplasm with frequent cross striations and intracytoplasmic rod-like inclusions. There were admixed well-differentiated skeletal muscle cells. The

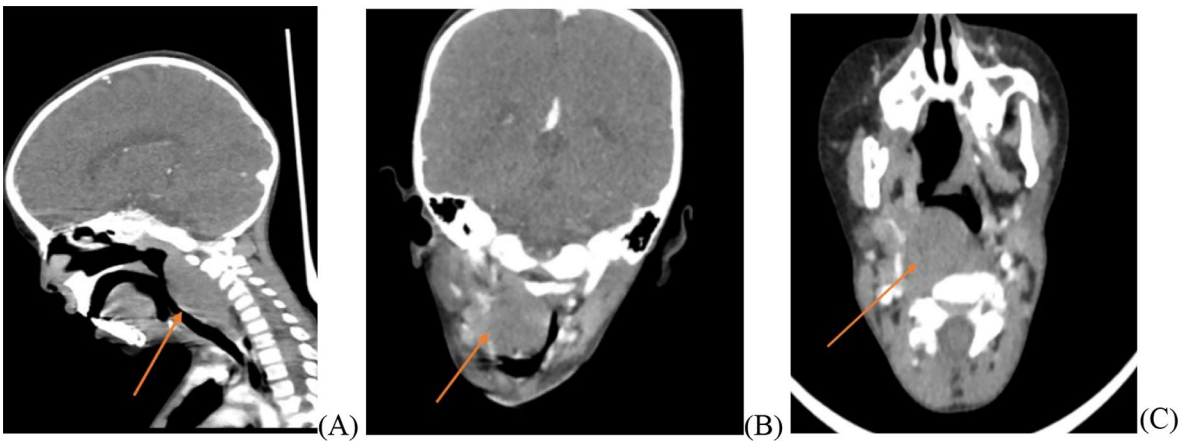
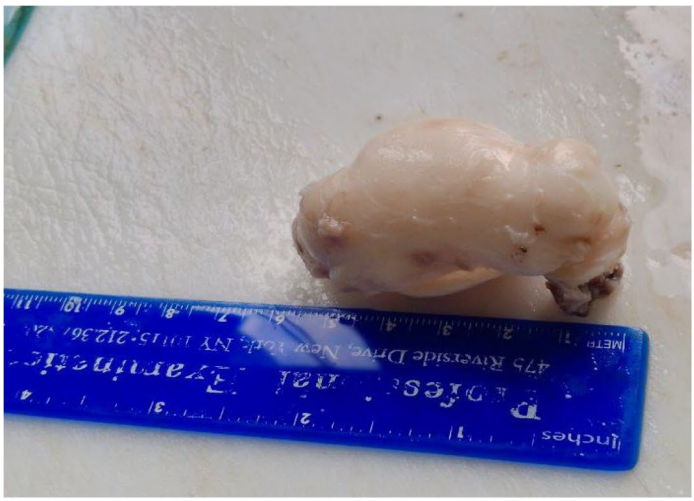
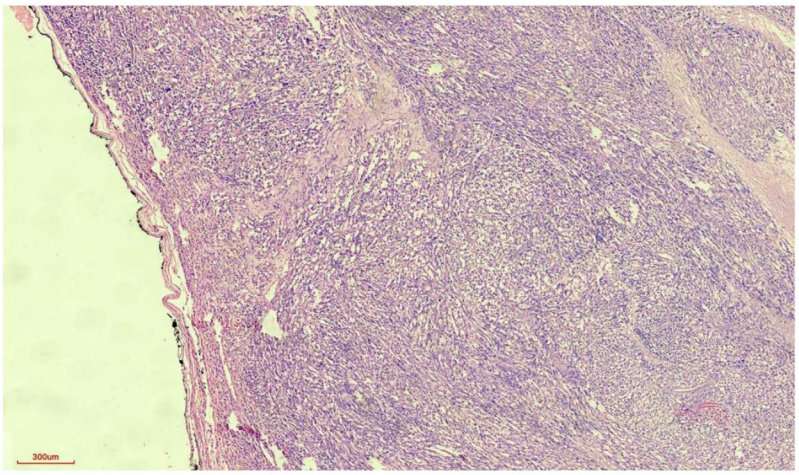


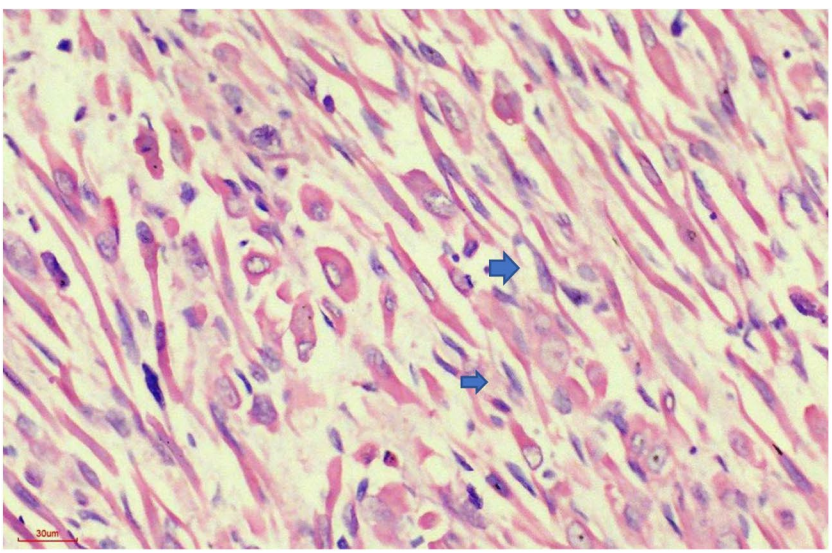
Fig. 1 post-contrast neck Ct images sagittal **A** coronal **B** and axial **C**, All the selected images demonstrate soft tissue mass that has homogenous attenuation at retropharyngeal space (orange arrow) with no significant calcification, necrosis, or adjacent bone destruction



A grey white well-circumscribed mass



B *Well-circumscribed periphery (40 x magnification)*



C Fascicular arrangement of strap cells, (arrows) 400 x magnification

Fig. 2 **A** grey white well-circumscribed mass. **B**: Well-circumscribed periphery (40×magnification). **C**: Fascicular arrangement of strap cells, (arrows) 400×magnification

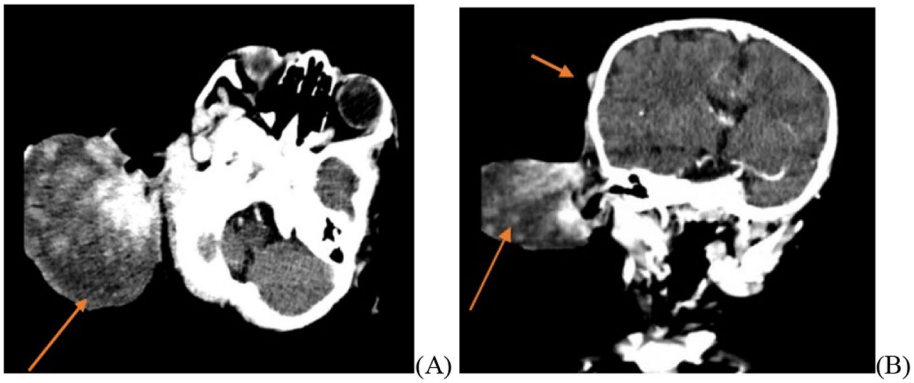


Fig. 3 Post-contrast axial **A** and coronal **B** brain CT, both images demonstrate well-demarcated soft tissue mass located at the mastoid area that has increased central flow with no bone destruction or extension to intracranial space (orange arrow). **A** small scalp lesion is noted that has no imaging appearance of aggressive (**B** -small arrow)

nuclei were small, round, and vesicular, and some with prominent nucleoli with no mitotic activity or atypia. Immunohistochemistry revealed positivity for muscle-specific actin, Desmin, and myoglobulin. The biopsy also revealed a subcutaneous variable-sized ecstatic thin-walled, and thick-walled vessels set in a fibrous stroma with plump spindle to stellate cells with scattered inflammatory cells. There were fascicles of slender cells with tapering eosinophilic processes (skeletal muscle fiber) and rhabdomoblasts with no mitosis, necrosis, or atypia (Fig. 4A–D). Sections through the lymph nodes and salivary glands were unremarkable. The child is doing well and is on her 18 month post-surgery follow-up with no recurrence.

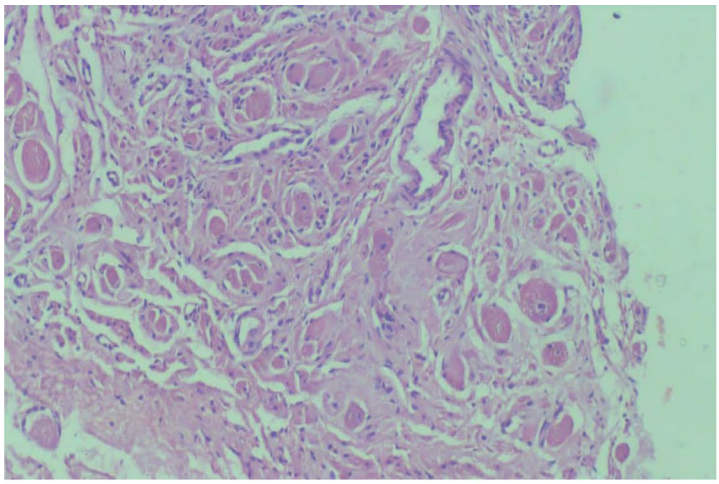
2.3 Case three

An 8-year-old male child presented with crampy abdominal pain of six months duration with infrequent vomiting episodes. Abdominal ultrasound showed a celiac Axis region ill-defined heterogeneous mass lesion, and a post-contrast abdominal CT scan showed a 2.3 cm × 2.3 cm × 1.8 cm well-defined homogenously enhancing lesion seen in the midline of the upper retroperitoneum (Fig. 5A–D). The lesion displaced the pancreatic head anteriorly and insinuated between the aorta and pancreatic head. The Celiac trunk and its common hepatic and proximal splenic artery seemed to be encased with no luminal narrowing. There was no internal calcification or necrosis seen. There was a clear fat plane between the mass and adjacent organs (the pancreatic head anteriorly, the aorta posteriorly, the liver, and the stomach laterally). There were no associated retroperitoneal lymph nodes seen. The liver, spleen, pancreas, bilateral kidneys, bowels, and lung bases were normal. With the clinical presentation and radiological finding neuroblastic tumors and rhabdomyosarcoma were considered as differential diagnoses, and biopsy was recommended to confirm the diagnosis. The patient underwent exploratory laparotomy and mass excision. The biopsy showed tissue fragments with the loose proliferation of spindle cells having short ovoid nuclei interspersed by abundant large rhabdoid cells with cytoplasmic striations, and admixed areas with small round to stellate cells with absent mitosis suggesting rhabdomyoma. The immunohistochemistry custom panel showed a Ki-67 index of less than 3%, smooth muscle actin was diffuse and strongly positive (85%), and myogenin positivity was less than 5%. The child is doing well and is on his second year post-surgery follow-up with no recurrence.

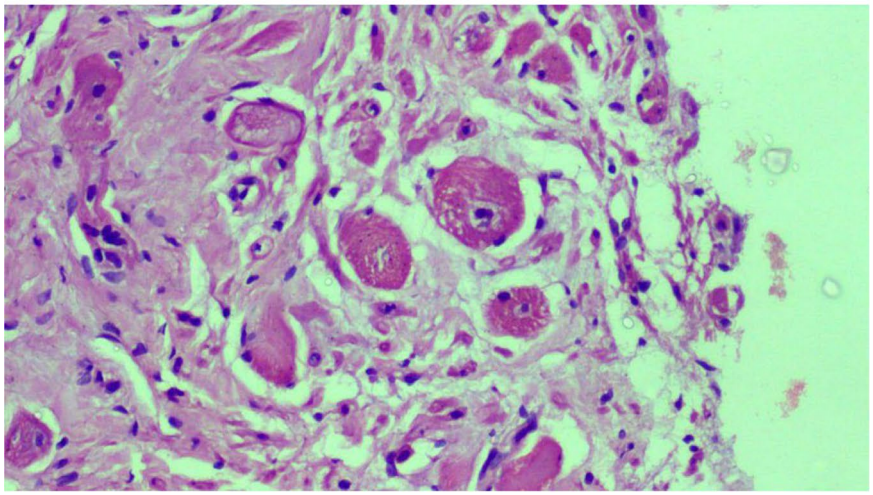
3 Discussion

Rhabdomyoma is an extremely rare benign tumor of the soft tissue of skeletal muscle with mature skeletal muscle differentiation, which can be classified into cardiac or extracardiac [1, 2]. The extra-cardiac rhabdomyoma can be divided into fetal, genital, and adult subtypes [3, 4].The fetal rhabdomyoma subtype arises in striated muscle, particularly in children under 3 years of age and with a predilection to the head and neck region [5, 6]. A few cases of extracardiac rhabdomyoma have been reported, but rhabdomyoma in the retroperitoneum and para-pharyngeal area is an infrequent phenomenon in clinical practice [6–8]. Surgical excision has been the mainstay of treatment and local recurrence has been reported due to incomplete surgical excision [9–12].

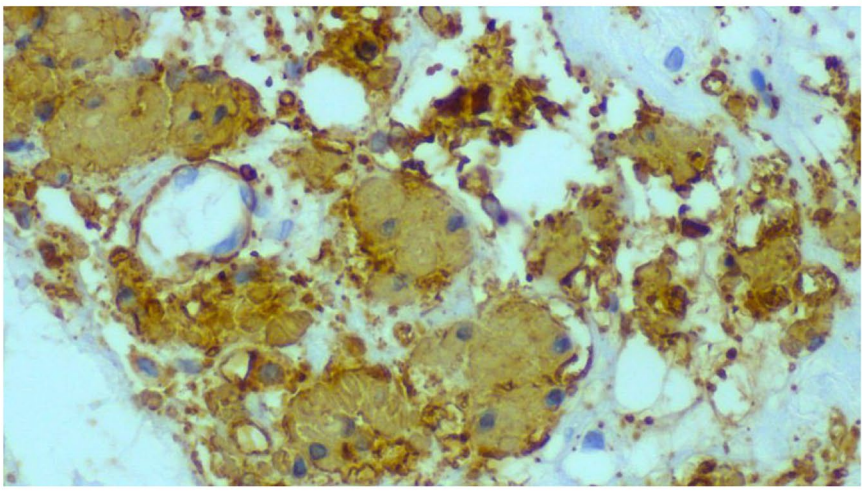
Malignant transformation of rhabdomyoma is very uncommon, though there are a few case reports in the literature [13]. In our case, the first case was para-pharyngeal rhabdomyoma, she had been treated with transcervical excision of the mass and she had no local recurrence on follow-up. The second child has a posterior auricular rhabdomyoma and the



A 10x objective microscopy showing spindle cells with ovoid nuclei interspersed with rhabdoid cells



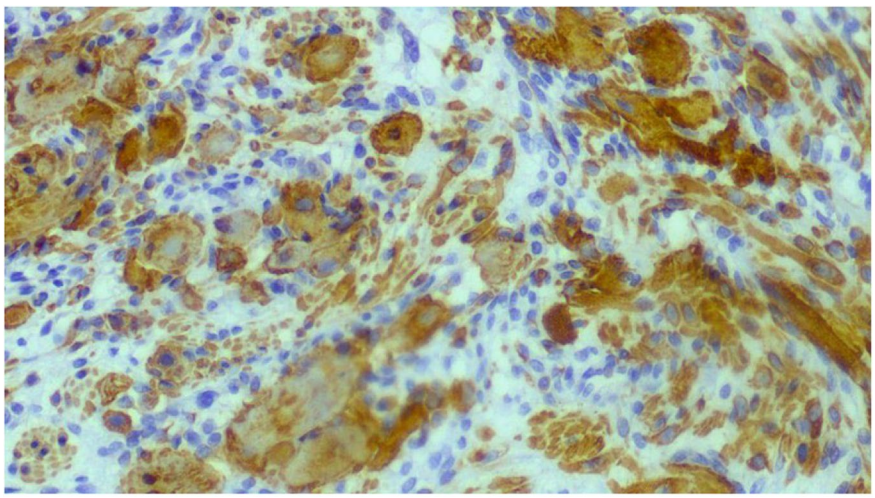
B 40x objective, Hematoxylin and Eosin section, large neoplastic cells with pinkish fibrillary cytoplasm and small nuclei.



C Immunohistochemistry: Muclе-specific actin positive in the large polygonal cells

Fig. 4 10 × objective microscopy showing spindle cells with ovoid nuclei interspersed with rhabdoid cells. **B**: 40 × objective, Hematoxylin and Eosin section, large neoplastic cells with pinkish fibrillary cytoplasm and small nuclei. **C**: Immunohistochemistry: Muscle-specific actin positive in the large polygonal cells. **D**: Immunohistochemistry showed Desmin diffuse expression

Fig. 4 (continued)



D Immunohistochemistry showed Desmin diffuse expression

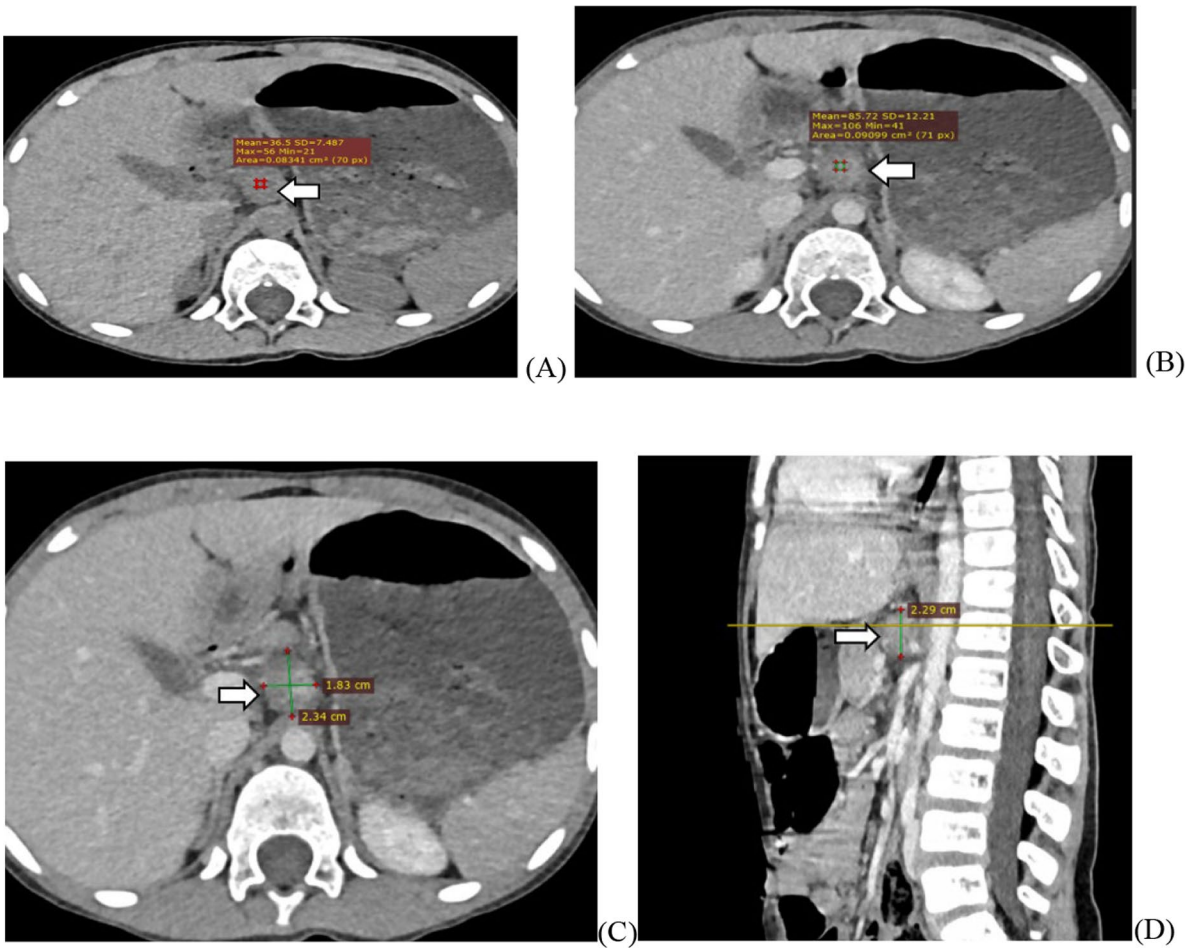


Fig. 5 **A**–**D**: Upper retroperitoneal well-defined mass (arrow) A-pre-contrast image and B-post-contrast image showing homogenous contrast enhancement. C-axial and D- sagittal image showing a well-defined homogenously enhancing midline upper retroperitoneal mass measuring 2.3 cm × 2.3 cm × 1.8 cm in size

tumor was excised completely, the child is doing well on follow-up. The third child had a biopsy-proven retroperitoneum rhabdomyoma, and he is doing well on his second year post-surgery follow-up with no recurrence.

Although rhabdomyoma is a very rare tumor entity, clinicians should be aware of the existence of this benign tumor, and the possibility of rhabdomyoma should be in the differential diagnosis of soft tissue tumors [14]. Pre-operative diagnosis based solely on clinical presentation and imaging is challenging. Tissue diagnosis is important to rule other potential malignant tumors such as soft tissue sarcoma, and neuroblastic tumors.

In this report, we present three cases of rhabdomyoma originating from the pharynx, retroperitoneum, and post-auricular regions. Pre-operative diagnosis based solely on clinical presentation and imaging is challenging and histological tissue confirmation is crucial to diagnosis rhadomyoma. Surgery is the standard treatment modality for extra-cardiac rhabdomyoma, and local recurrence is rare after complete excision.

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Author contributions G.A., A.M, G.F., M.N.,T.M and T.N.: Inception of the idea, data collection, data analysis, write-up, and editing of the manuscript; A.Z, F.O, and Y.A., Data analysis, review of the manuscript and prepared radiological Figs. 1A–C, 3A–B, and 5A-D. A.T., A.D., and M.A. wrote up the pathology and prepared Figs. 2A–C, and 4A-D. All authors reviewed the manuscript and approved the final version of the manuscript for publication.

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Data availability The corresponding author can provide the data sets used in this study upon a reasonable request.

Declarations

Ethics approval and consent to participate The study was approved by the Addis Ababa University, Department's of Pediatrics Research and Ethics Committee and Institutional Review Board, and the study was conducted in accordance with the WMA Declaration of Helsinki.

Consent for publication Written informed consent was obtained from the patient's parents for anonymized patient information to be published in this article**.**

Competing interests The authors declare no competing interests.

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