TPM Vol. 32, No. S3, 2025
ISSN: 1972-6325
https://www.tpmap.org/

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UNCOMMON AURAL MASS: SPINDLE CELL VARIANT OF EMBRYONAL RHABDOMYOSARCOMA UNVEILED - A RARE CASE REPORT

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Abstract:
Rhabdomyosarcoma is most common soft tissue sarcoma often affects children arising from skeletal muscle. A 4-year-old girl presented with 2weeks history of left ear blood-stained purulent discharge and aural growth. CE-MRI revealed a homogeneous soft tissue lesion in the left external auditory canal extending into the middle ear and mastoid air cells. HPE confirmed the spindle cell variant of Embryonal subtype RMS, with MyoD1 and Ki67 positivity, involving the left mastoid, and treated with neoadjuvant chemotherapy, radical mastoidectomy, radiotherapy, and additional chemotherapy.This aggressive multimodal approach achieved favorable initial outcomes,with no recurrence,emphasizingearly,intensive therapy's role in localized disease control and prognosis improvement.

Keywords: Embryonal Rhabdomyosarcoma ,Chronic suppurative otitis media, MyoD1 mutation, Multimodal aproach, Intergroup Rhabdomyosarcoma study

INTRODUCTION

Rhabdomyosarcoma (RMS), arising from poorly differentiated mesenchymal cells, represents 3% to 4.5% of pediatric malignancies, making it the most common soft-tissue sarcoma in children. This malignancy is particularly significant due to its aggressive nature and the challenges it poses in diagnosis and treatment.[1,2] Most RMS cases in the head and neck region involve the orbit(33%), oral cavity(29%), face and neck (24%), ear and temporal bone involvement is rare. These parameningeal sites are critical because their involvement often leads to delayed diagnoses due to the nonspecific symptoms that mimic more benign conditions, such as chronic suppurative otitis media.[1,3] Sarcomas in the temporal bone, including the middle ear and mastoid accounts for less than 5% of temporal bone malignancies. Rhabdomyosarcoma manifests primarily in four histological variants: Embryonal, Alveolar, Botryoidal, and Pleomorphic. The Embryonal type, constituting 70% of RMS cases, is the most prevalent and generally carries a better prognosis than the alveolar type, which is associated with a poorer outcome. The significance of this case report lies in its contribution to the limited pool of literature on RMS of the temporal bone, particularly in young children.[4] This report underscores the importance of early recognition and intervention with chemoradiotherapy , which are critical for enhancing survival rates and quality of life in affected children.

Case Report:
A 4-year-old girl who initially presented with a 2 weeks history of a growth inside her left ear accompanied by blood-stained purulent discharge with hearing loss and not associated with ear pain and facial weakness. No other significant ENT complaints. Clinical examination revealed a reddish, polypoidal, fragile mass resembles granulation tissue within the left external auditory canal (EAC) that was non-bleeding on touch, not able to probe around the margins (figure 1A) and was not associated with palpable neck nodes or cranial nerve symptoms. Pure tone audiometry revealed left sided moderate conductive hearing loss.
CE-MRI of the head showed a homogeneous soft tissue lesion with diffusion restriction located in the left EAC, extending into the middle ear and the bony part of the Eustachian tube (figure 2).There was no evidence of ossicular chain erosion or involvement of the inner ear structures, indicating a less aggressive disease or early stage.
A biopsy was done from the polypoidal tissues, showed stratified squamous epithelial lining, spindle and oval cells with eosinophilic cytoplasm and mild to moderate nuclear pleomorphism (figure 1B-D). The stroma appeared loose and myxoid, containing scattered thin-walled blood vessels. Crucially, Rhabdomyoblastic differentiation and mitotic activity were observed without necrosis, which suggested a diagnosis of spindle cell sarcoma. Immunohistochemical staining was definitive, showing intense nuclear positivity for MyoD1 and an increased Ki67 index, which are indicative of Spindle cell variant of Embryonal RMS.
The staging of the tumor was determined as Stage II, characterized by its unfavourable site, a size less than 5 cm (T1), no nodal involvement (N0), and no metastasis (M0). The treatment strategy comprised a multidisciplinary approach starting with neoadjuvant chemotherapy using vincristine, actinomycin, and cyclophosphamide administered over two months with 4 cycles. This was followed by a radical mastoidectomy performed through a postauricular approach, indicating gross resection of regional disease without lymph node involvement. No postoperative complications noted , postoperative management included radiotherapy dose of 36 Gy at 1.8Gy per fraction in 20 fractions, 5 fractions per week, and further 6 cycles of chemotherapy to consolidate the treatment gains and mitigate any risk of recurrence. Patient was followed up for 1 year and PET-CT done showed no recurrence or residual tumor.

DISCUSSION

Embryonal rhabdomyosarcoma (RMS) of the temporal bone is an exceedingly rare and aggressive malignancy, particularly in the pediatric population.[5] Rhabdomyosarcomas (RMS) shows a bimodal age distribution, peaking at 2–5 years and during adolescence, with 63% of cases in children under 10. Slightly more common in males and Caucasians, RMS is the third most frequent childhood tumor after neuroblastoma and nephroblastoma, often affecting the nasopharynx, parapharyngeal space, pterygopalatine fossa, inner ear, urogenital tract, and digestive system. Embryonal RMS is most common RMS but invovlement of temporal bone is rare and is characteristically associated with the loss of heterozygosity at a specific locus on the short arm of chromosome 11 (11p15). Alveolar rhabdomyosarcoma (RMS) involves translocations between the FKHR gene on chromosome 13q and PAX family genes on chromosome 2 (PAX3) or chromosome 1 (PAX7). TP53 mutations occur in both embryonal and alveolar RMS, while elevated N-myc expression (10% in alveolar RMS) and N-ras/K-ras mutations are more common in embryonal RMS.[6]
Based on clinical evaluation ,the differential diagnosis encompasses benign tumors such as hemangioma, meningioma, pilomatrixoma, glomus tumor, and lymphangioma, as well as malignant tumors including juvenile fibromatosis, chondrosarcoma, lymphoma, and neurofibroma. The WHO (2020) classifies Rhabdomyosarcoma into Embryonal, Alveolar, Spindle Cell (including newer variants), and Pleomorphic types. Prognosis varies by subtype: Botryoid and Spindle Cell RMS have the best outcomes, Embryonal RMS has an intermediate prognosis, while Alveolar and Undifferentiated RMS are associated with poor outcomes. Recent studies have refined RMS classification with the identification of three new subtypes: RMS with MYOD1 mutations, TFCP2 fusions, and VGLL2/NCOA2 fusions. Parameningeal Rhabdomyosarcoma (RMS) with intracranial extension can affect the facial, oculomotor, trochlear, and abducens nerve, resulting in diplopia.[6,9]
Immunohistochemistry is essential for distinguishing RMS from other small round cell neoplasms like lymphoma (CD20, CD3 positive) and Ewing's sarcoma/PNET (CD99 positive), as RMS lacks these markers. RMS cells express Desmin, Myogenin, CD56, muscle-specific actin, Myoglobin, Vimentin, and MyoD1. Fetalhemoglobin detection via immunoperoxidase staining is a reliable diagnostic marker for RMS.[6] In our case it is MyoD1 high positivity.
Staging for RMS includes: Stage I includes tumors at favorable sites, while Stage II involves small tumors (<5 cm) at unfavorable sites without lymph node involvement (N_0, Nx). Stage III comprises large tumors (≥5 cm) or those with lymph node involvement (N_1) at unfavorable sites. Stage IV indicates clinically evident metastatic disease.The Intergroup Rhabdomyosarcoma Study (IRS) protocols, introduced in 1972 and updated through IRS

II (1978), IRS III (1984), and IRS IV (1991), transformed RMS treatment with multi-agent chemoradiation, reserving surgery for cases without significant morbidity. New agents like etoposide, ifosfamide, and melphalan were added to the standard regimen of vincristine, actinomycin-D, and cyclophosphamide, enhancing efficacy. Radiotherapy, particularly for head and neck tumors, is administered at 4000–4500 Gy, optimizing treatment while reducing toxicity. Brachytherapy shows promise in head and neck rhabdomyosarcoma with effective control and minimal side effects. These protocols have significantly improved outcomes in rhabdomyosarcoma management.[10] Neoadjuvant chemotherapy is vital in treating RMS by shrinking tumors and enabling complete surgical resection, as seen in this case. This reduction allowed for clear-margin resection during radical mastoidectomy. Studies consistently shows chemotherapy's role in improving surgical outcomes, while postoperative radiotherapy targets residual microscopic disease, crucial for preventing recurrence. Adjuvant chemotherapy addresses potential micro-metastases, contributing to better survival rates. Advances in multimodal therapy have significantly improved prognosis, with 5-year survival rates for localized RMS rising to 74–77% and disease-free survival ranging from 58–74%. However, salvage therapy for recurrent RMS remains challenging. Early detection and aggressive treatment are key to these improved outcomes.[7,8]

CONCLUSION

Spindle cell variant of Embryonal Rhadomyosarcoma is uncommon and may present with facial paralysis, but advances in chemotherapy and radiotherapy achieve high remission rates and preserve quality of life. while annual PET-CT scans aid in early recurrence detection and prompt treatment. Early diagnosis through imaging and histopathology, combined with a robust multimodal treatment approach, is vital for improving outcomes in rare and aggressive tumors like Embryonal RMS. This case underscores the need for heightened clinical suspicion and timely intervention to enhance survival and quality of life for affected patients.

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Figures legends

Figure 1: Gross clinical and histopathological feature of the left EAC growth

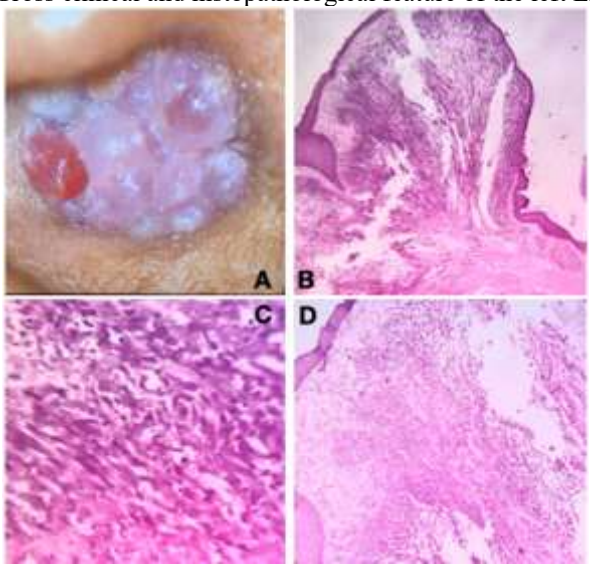

Figure 2: Imaging studies (CE-MRI) showing a homogeneous soft tissue lesion with diffusion restriction