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Case Report

Rare Presentations of Embryonal Rhabdomyosarcoma in the Middle Ear of a 3-Year-Old Boy and Girl.

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Abstract

Embryonal rhabdomyosarcoma is a distinct subtype of rhabdomyosarcoma commonly seen in children less than 5 years of age, but it can appear at any age. It's frequently seen at the head and neck region, but rare sites include the ear. It often presents with facial nerve palsy. We present two extraordinary cases of embryonal rhabdomyosarcoma in the middle ear in a 3-year-old boy and girl.

Keywords: Embryonal Rhabdomyosarcoma; Middle Ear; 3-Year-Old; Boy; Girl.

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Introduction

Rhabdomyosarcomas (RMS) are soft tissue malignancies that display rhabdomyoblastic differentiation.[1] They are heterogeneous and are divided into 4 distinctive subtypes according to the 2013 WHO Classification of Soft Tissue and Bone Tumours – embryonal rhabdomyosarcoma (ERMS), alveolar rhabdomyosarcoma (ARMS), pleomorphic rhabdomyosarcoma (PRMS) and spindle cell/sclerosing rhabdomyosarcoma (ScRMS).[2] However, three new subtypes – RMS with MYOD1 mutation, RMS with VGLL2/NCOA2 mutation and RMS with TFCP2 mutation – have emerged based on clinicopathologic and molecular features.[1] Embryonal RMS is the most common subtype frequently seen in children, usually boys and between 1 to 4 years of age, but can occur at any age and with a penchant for the head and neck.[2] Other frequent sites are the orbit, pharynx and mouth, with the ear and temporal bones as infrequent sites.[3] Rhabdomyosarcoma of the ear usually mimics chronic suppurative otitis media, making diagnosis challenging.[4] Diagnostic modalities include computed tomographic (CT) scan and/or magnetic resonance imaging (MRI), and lesional biopsy. Treatment is multimodal and includes surgery, chemo, molecular-targeted-, therapy, immune-, and radiation therapies.[3]

Case History

Case 1: A 3-year-old boy was admitted to the emergency paediatric unit on account of fever, right eye swelling and protrusion with inability to close the right eye six weeks before presentation. There was an associated history of deviation of the mouth to the left. However, there were no ear symptoms, nasal or throat symptoms and no convulsions or loss of consciousness. At the onset of symptoms, he was taken to a comprehensive health care centre where he was given oral medications, after which the eye swelling and protrusion resolved. He, however, started having right ear discharge with swelling around the right ear 5 weeks after the onset of the initial symptoms. A diagnosis of Chronic suppurative otitis media, complicated by mastoiditis, facial nerve palsy and right orbital cellulitis was made by the paediatricians. However, a review by the Ear, Nose and Throat (ENT) surgeons revealed a normally shaped and sized pinna, a swollen right peri-auricular area, and the rightexternal auditory canal was completely blocked by a fleshy growth, preventing the view of the right tympanic membrane. However, the left tympanic membrane was intact and shiny, with left-sided deviation of the mouth and loss of the right nasolabial fold. The right eye was mildly swollen with mild discharge and greater than one-thirdof corneal scarring. A provisional diagnosis of a right ear tumour was made. A computed tomographic (CT) scan was requested but was not done due to financial constraints. A biopsy was taken and sent to the pathology laboratory for analysis.

Grossly, three fragments of greyish-white tissue aggregating 1x1cm across were received. Microscopy showed a circumscribed lesion lined by keratinizing stratified squamous epithelium overlying peripheral circumferential extensive areas of necro-inflammation, within which were alternating hypercellular and pauci-cellular areas thatwere composed of infiltrating sheets of atypical round, oval and spindly cells having hyperchromatic nuclei and scanty cytoplasm disposed within a myxoid stroma demonstrating numerous variable - sized vascular channels

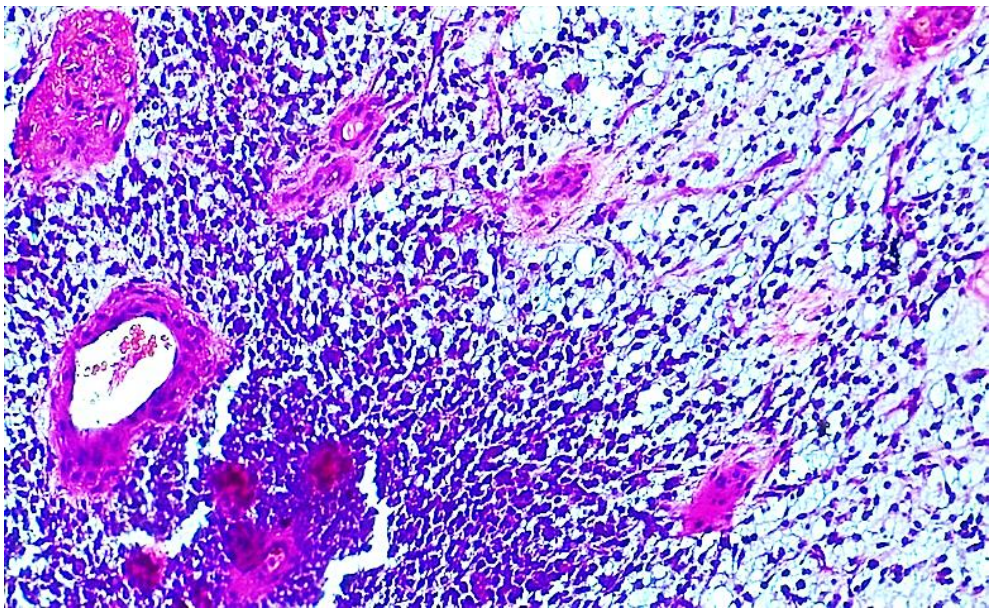

Figure 1: The histological (H&E) section shows alternating hypercellular and pauci-cellular areas composed of infiltrating sheets of atypical cells within a myxoid stroma exhibiting numerous variable-sized vascular channels, X10.

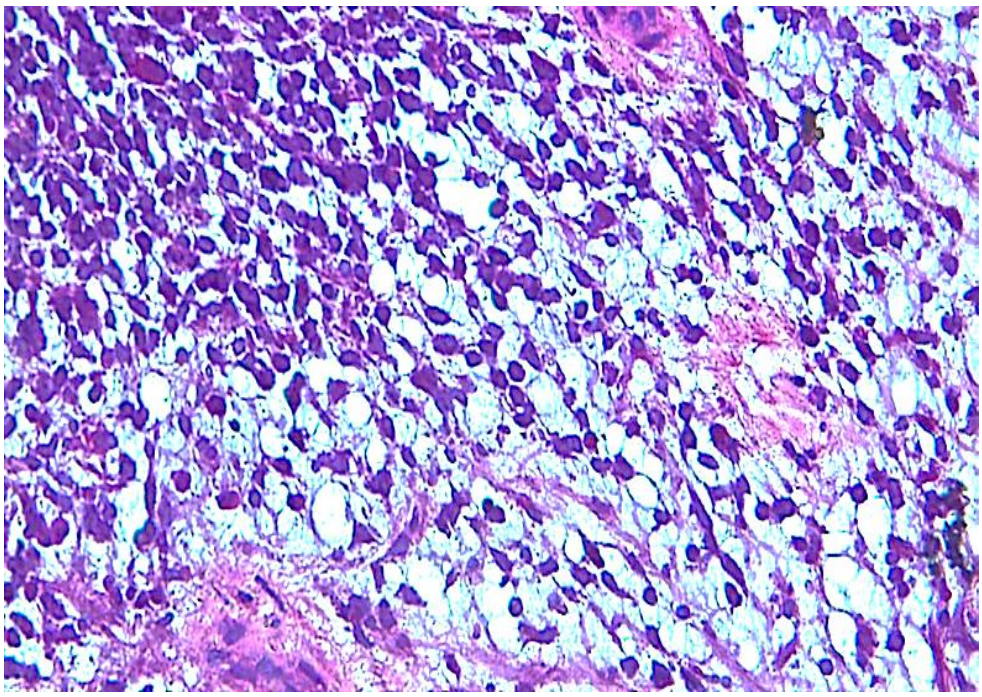

Figure 2: The histological section shows infiltrating sheets of atypical round, oval and spindly cells having hyperchromatic nuclei and scanty cytoplasm disposed within a myxoid stroma, X40.

The atypical cells were focally positive for desmin and negative for S100 immunohistochemistry markers **[Figures 3 & 4]**. A diagnosis of embryonal rhabdomyosarcoma was made. The patient was lost to follow-up.

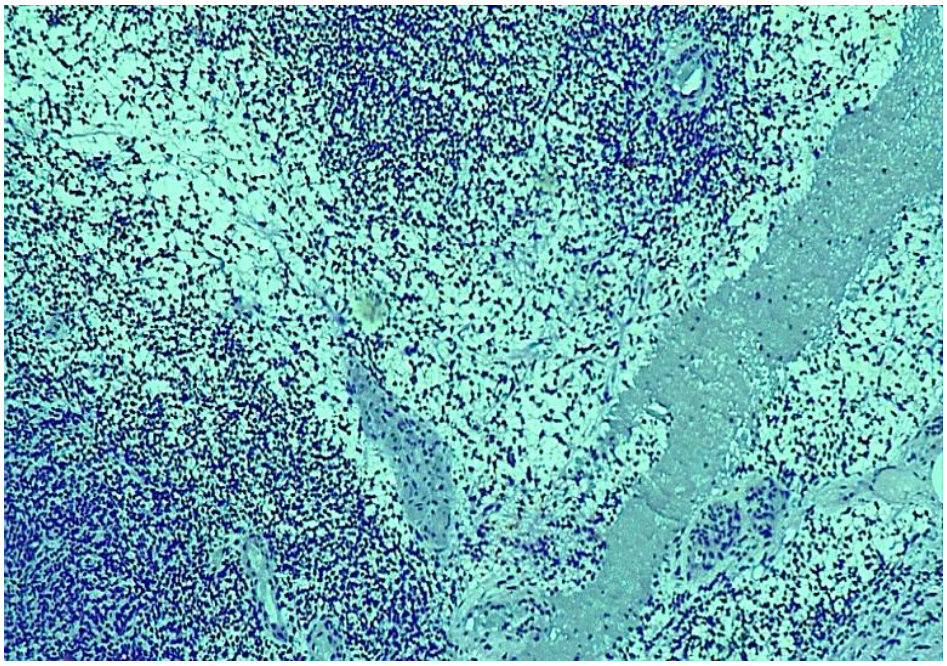

Figure 3: Photomicrograph showing negative S100 immunostaining, X100.

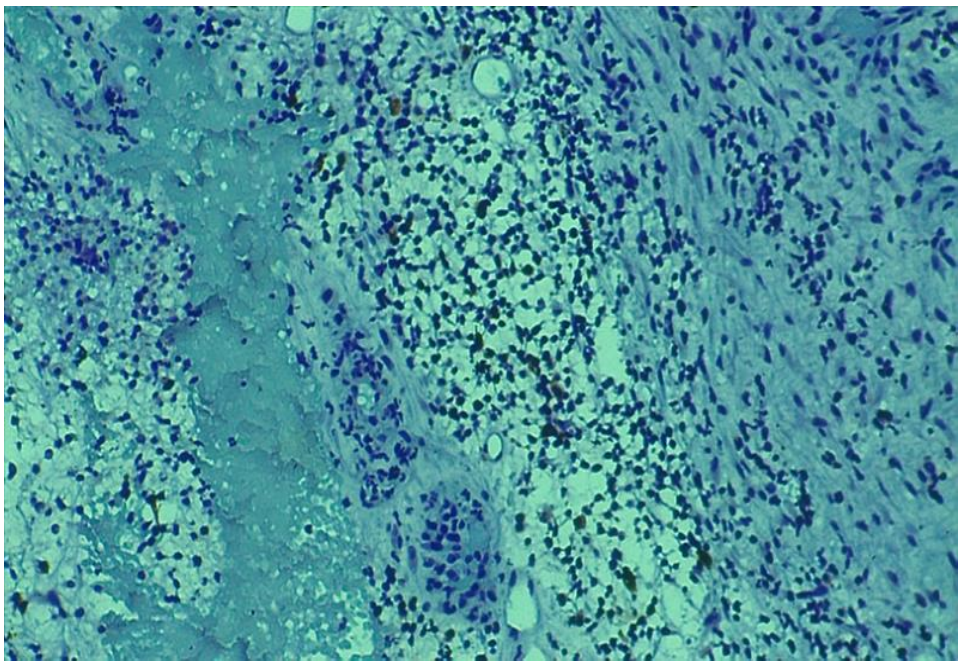

Figure 4: Photomicrograph showing embryonal rhabdomyosarcoma demonstrating focally positive desmin immunostaining, X10.

Case 2: A 3-year-old girl presented to the emergency paediatric unit with swelling of the right ear and deviation of the mouth to the left side 6 weeks before presentation. They subsequently noticed a protrusion of a whitish substance through the right ear with swelling around the ear, estimated to be around the size of her fist. It then progressed over these 6 weeks to its current size, quadruple the size of her fist. Since the onset, no regression has been noticed. There was an associated history of headache and occasional convulsion (twice) but no history of loss of consciousness. There were no associated swellings on any other part of the body.

At the onset of symptoms, the patient was taken to a traditional healer, who made an incision and reportedly removed some flesh tissue while applying traditional concoctions. There was no relief in symptoms, prompting the patient to be taken to a peripheral hospital, where some medications were given. However, the patient continued to deteriorate, necessitating presentation to this facility. An assessment of a left facial tumour with intracranial extension, subdural collection and necrotizing fascitis was made. A review was conducted by plastic and neurosurgeons. Physical examination revealed facial asymmetry, with a deviation of the mouth's angle to the left, a squint in the left eye, and an irregular, large, firm mass protruding from the right ear. The mass measured 8x10cm and exhibited multiple ulcerations in the post-auricular and temporal regions, the largest being about 5cm in diameter in the post-auricular region, which had a central area of necrosis and purulent discharge. A provisional diagnosis of rhabdomyosarcoma with intracranial extension was made.

A computed tomographic (CT) scan was requested and showed a heterogeneously enhancing mass (HU: 20-59) with lobulated margins in the parotid gland's region and the ear's adjacent soft tissue on the right side. It measured 99 x 79 x 77mm in the longitudinal, transverse and anteroposterior dimensions. The right inner, middle and outer ear were destroyed and invaded by the mass with lytic destructions of the temporal bone, sphenoid and clivus, sparing only the pinna. Part of this mass was also seen in the temporal and parietal lobes with associated moderate right subdural collection (HU: 9-15). However, the remaining parts of the brain appeared within normal limits. A biopsy was taken and sent to the pathology laboratory for analysis.

Grossly, three greyish whites to tan tissue fragments each measuring 2x1x1cm were received. Microscopy showed sheets and fascicles of atypical primitive mesenchymal cells. The cells were oval to spindle to round with hyperchromatic nuclei and eosinophilic cytoplasm. Cells having eccentric hyperchromatic round nuclei with abundant eosinophilic cytoplasm were also seen. These were disposed within a fibromyxoid stroma exhibiting moderate necrosis **[Figures 5 & 6]**. The atypical cells were negative for desmin and S100 immunohistochemistry markers **[Figures 7 & 8].** A diagnosis of Embryonal Rhabdomyosarcoma was made. The patient was lost to follow-up.

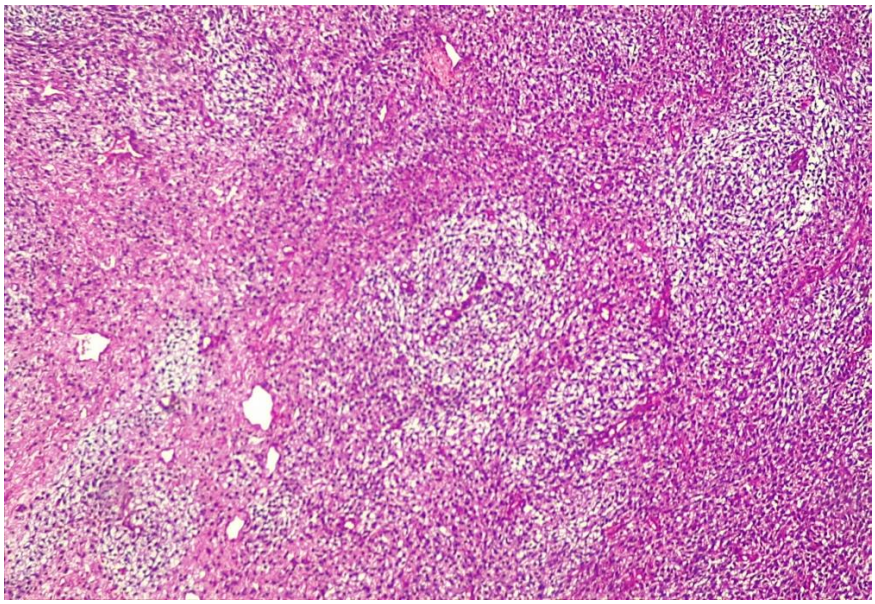

Figure 5: Photomicrograph of H&E-stained section showing alternating hypercellular and paucicellular areas composed of infiltrating sheets of atypical cells disposed within a myxoid stroma exhibiting variable-sized vascular channels, X10.

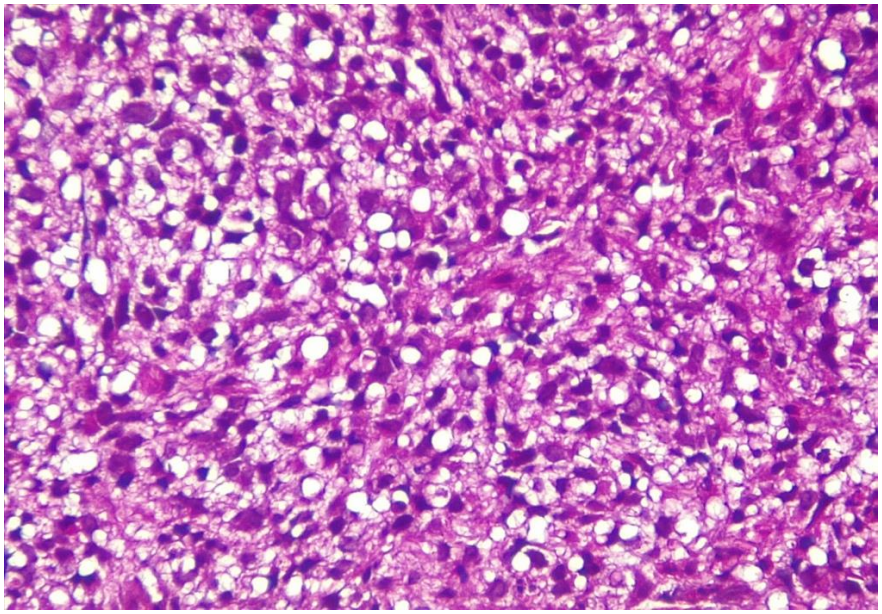

Figure 6: Photomicrograph of H&E-stained section showing a tumour composed of infiltrating sheets of atypical round to oval and spindled cells that have hyperchromatic nuclei and scanty cytoplasm disposed within a myxoid stroma, X40.

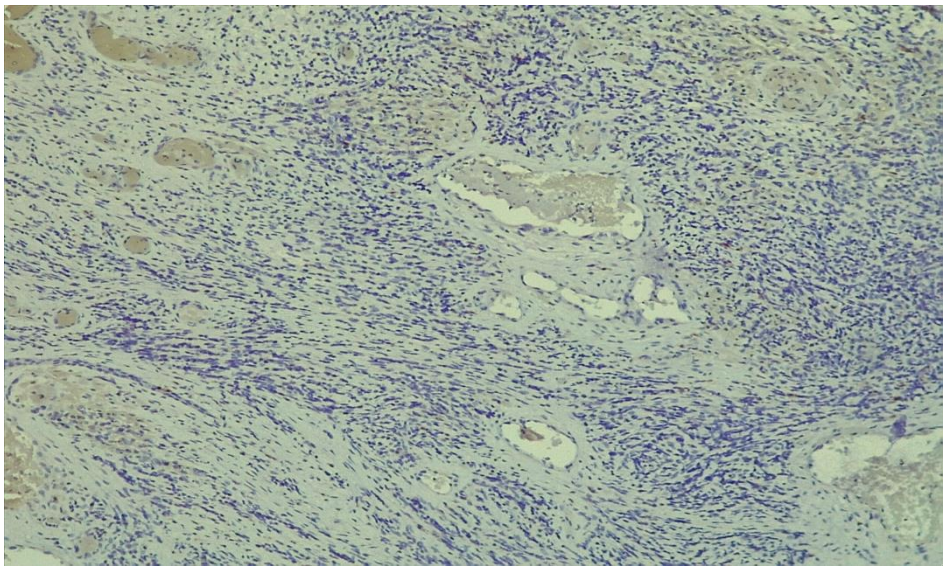

Figure 7: Photomicrograph showing embryonal rhabdomyosarcoma demonstrating negative S100 immunostaining, X10.

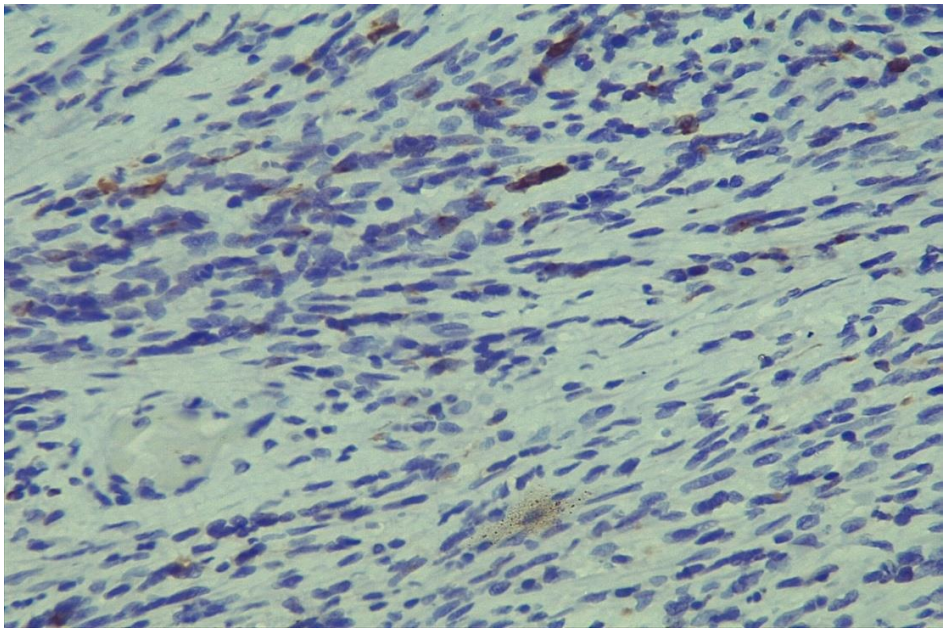

Figure 8: Photomicrograph showing embryonal rhabdomyosarcoma demonstrating negative desmin immunostaining, X10.

Discussion

Rhabdomyosarcoma is the most common soft tissue sarcoma in children with the embryonal subtype as the commonest entity.[5] It is frequently seen in children within 5 years of age but can occur at any age and it usually has a male predilection.[6]This is similar to what is seen in the index study whereby the patients are both 3 years old with one being a boy and the other a girl. Similar studies in 3- to 8-year-old boys were seen in India,[7, 8] the United Kingdom,[9] and Saudi Arabia;[10] and cases in 2- and 5-year-old girls in Pakistan and India respectively.[4, 11]

The most frequent sites of ERMS are the head and neck, and the genito-urinary tract. However, the ear and temporal bones are uncommon sites.[3] In this study, the lesion was seen within the right and left ear cavities with lytic destruction of the temporal bones, sphenoid and clivus. This is similar to findings from a study in Iran of an ERMS in the mastoid and middle ear in a 3-year-old girl.[4]In contrast, a study by Russo et al. in Italy reported ERMS on the head, limbs and skin of a newborn baby girl.[12] Again, ERMS of the tongue was reported in a 46-year-old man in Madrid, Spain, and a case of laryngeal ERMS was reported in China.[13, 14]

Embryonal rhabdomyosarcoma should always be a strong differential in any child presenting with swelling in the tympanic cavity, ear discharge, and orbital swelling.[14] These symptoms are similar to what is seen in this index case.A similar presentation was reported by Kumar in 2 cases reported.[15]Head and neck rhabdomyosarcoma may also present with cranial nerve palsy at the initial presentation.[16]This report is similar to the findings in our study where both patients presented with facial nerve palsy. At a certain stage, ERMS may extend into the intracranial region, and the patient may present with features of space-occupying lesion.[17] A similar presentation was noted in one of our patients.

ERMS is diagnosed by histology however, histologic diagnosis usually presents with cellular heterogeneity and therefore, immunohistochemistry is needed for confirmation of the diagnosis.[17]A similar diagnostic modality was employed in confirming the histologic diagnoses of both cases in this report. Magnetic resonance imaging (MRI) and computed tomography (CT) scans are the radiologic diagnostic modalities of choice. MRI can delineate soft tissue and differentiate rhabdomyosarcoma from other soft tissue tumours while CT scan can determine bony invasion and relation with other contiguous structures.[18] CT scan was used in this report because MRI is not available in our centre. CT revealed features of bony destruction and extension into surrounding structures.

Conclusion

Embryonal rhabdomyosarcoma of the middle ear is an uncommon cancer. Though the prognosis is poor, early diagnosis and treatment, through the biopsy of the lesion, go a long way in alleviating patients' sufferings. A diagnosis of embryonal rhabdomyosarcoma should, therefore, be considered in all children presenting with otitis media, aural mass, and facial nerve palsy.

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