Embryonic rhabdomyosarcoma of the petrous bone in a child: a case report

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Abstract

Rhabdomyosarcoma (RMS) is a cancer of skeletal muscle origin, and the second most common soft tissue sarcoma encountered in childhood. The head and neck are common sites though the temporal bone is rare. Rhabdomyosarcoma represents 3.5% of all malignancies in children aged 0-14 years, with approximately 250 new cases diagnosed each year. Despite the more intensive management modalities including surgery and combination chemo-radiation, the outcome for patients with metastatic disease remains poor. Here, we report a case of temporal bone Embryonic RMS in a three and half year-old male who was seen at Mulago National Referral Hospital, Kampala in 2016 and describe the clinical, radiological and histopathological presentation of relevance to RMS.

Key Words: rhabdomyosarcoma, temporal bone, mesenchymal tissue, parameningeal, chronic suppurative otitis media, multi-modality therapy, Kampala

Introduction

Rhabdomyosarcoma (RMS) is a fast growing and highly malignant tumour of the soft tissue commonly affecting the head and neck.[1] It originates from mesenchymal tissue of highly myogenic differentiation of immature embryonic skeletal muscles.[2] This tumour accounts for 60% of sarcomas in children aged less than 5 years.[3, 1] RMS of the head and neck are divided anatomically into two based on their origin as parameningeal or non-parameningeal with the former having the worst prognosis.[5, 6] RMS of the temporal bone is very rare.[4] The predisposition to RMS is thought to be genetic mutation either in the form of chromosomal translocation or allelic loss which subsequently contributes to the oncogenic characteristic of the tumour.[2]

RMS has three known pathological types namely, embryonal rhabdomyosarcoma (85%), alveolar rhabdomyosarcoma (15%) and pleomorphic rhabdomyosarcoma (anaplastic rhabdomyosarcoma and undifferentiated sarcoma) with the former being common in children.[1, 8, 9]

RMS almost always mimics complicated chronic suppurative otitis media (CSOM) with ear discharge and ear canal mass. About 30% of patients have craniopathies at presentation. This may delay the diagnosis of RMS due to similarities in their clinical picture. The diagnosis of RMS is best confirmed through histopathology. Imaging is useful to determine the extent of disease.[11]

Multiple chemotherapy and radiation with or without tumour resection have greatly improved prognosis.[12] Ten percent of patients with RMS used to have a five years survival rate, but after the adoption of multi-modality therapy, outcomes were greatly improved.[11] Our three and half year-old male presented with features resembling a complicated CSOM, associated with facial weakness. The histopathology and immunostains of the ear polypoid mass were consistent with embryonal RMS.

Case presentation

A three and half year-old male was seen at Mulago National Referral Hospital,

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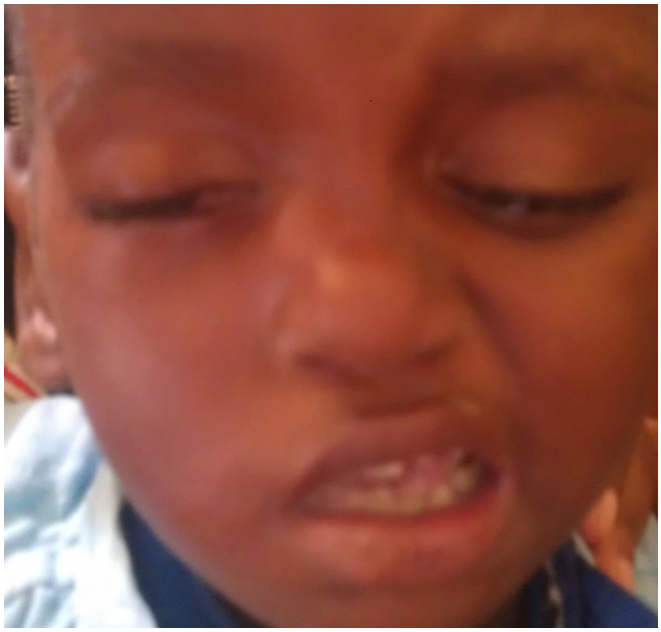


Figure 1. Right facial palsy and convergent squint of the right eye.

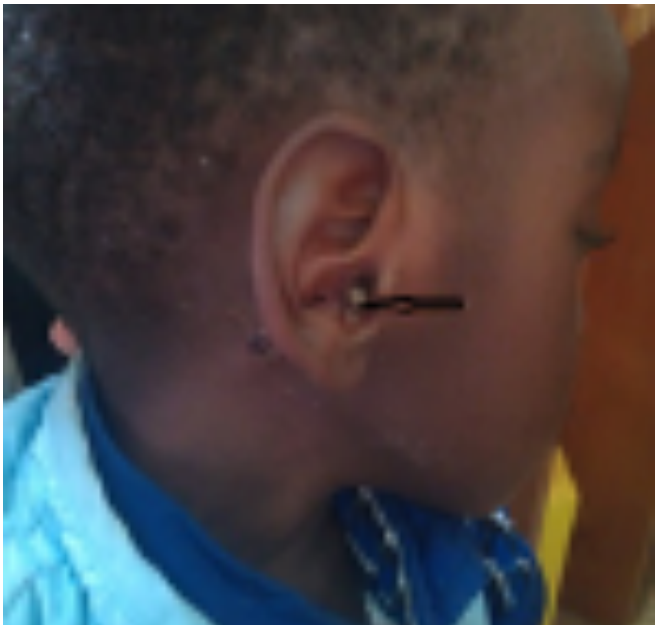


Figure 2. A polypoid aural mass seen occupying the whole right external ear canal.

Kampala in 2016. He presented with right ear discharge for a month, associated with swelling behind the ear and right facial weakness. The discharge was watery and blood stained later becoming purulent. He did not experience pain until a week prior to admission when the swelling behind the ear developed, associated with high fever. There were no convulsions. The mother noticed that the child's right eye did not close completely, with facial weakness, and associated drooling of saliva. There was no relevant past history, no history of trauma and no contact with others with respiratory symptoms.

Examination revealed a generally sick child. There was no palpable cervical lymphadenopathy, however the right facial palsy and convergent squint of the right eye were obvious (Figure 1).

The right auricle was displaced anteriorly and downward with a diffuse tendered post auricular swelling. The post-auricular crease was intact.

The external auditory canal (EAC) was full of debris which was sucked out revealing a pale, fleshy mass occupying the entire canal with a serosanguinous discharge. The tympanic membrane could not be seen (Figure 2).

A biopsy from the aural mass showed clusters of small round cells with hyper chromatic nuclei and eosinophilic cytoplasm separated by fibro vascular septae consistent with embryonic rhabdomyosarcoma. Immunohistochemistry found desmin, smooth muscle actin and myogenin which are considered to be of diagnostic value.

At first the suspicion was of complicated CSOM with an aural polyp. CT scan imaging is shown in Figures 3, 4a, 4b, 5 and 6.

The best option for this case was chemotherapy (cisplatin and vincristine every week for 4 cycles) and local control with radiotherapy in view of involvement of vital structures and the intracranial extension. Unfortunately, after three weeks from admission between diagnosis and the planned management, the child died most likely from the effects of metastases.

Discussion

In 1854 Weber described rhabdomyosarcoma (RMS) as a malignant neoplasm of soft tissue arising from skeletal muscle most commonly affecting the head and neck, genitourinary tract although other organs can be affected. [1] The incidence of RMS is more common in children between the ages of 1-4 years and accounts for 60% of soft tissue sarcomas in children.[3] It is considered to be the third commonest (35%) tumour in childhood after neuroblastoma and nephroblastoma with slight male preponderance.[4]

RMS of the head and neck is divided anatomically into two types: [6, 8]

1. Parameningeal- including the nose, nasopharynx, paranasal sinuses, mastoid region, infra-temporal, pterygopalatine fossae and middle ear.
2. Non-parameningeal- scalp, orbit, parotid gland, oral cavity, oropharynx and larynx

RMS of the ear or temporal bone is very rare. However, it commonly occurs in the head and neck region with the orbit constituting about one-third of cases, followed by oral cavity and pharynx (29%), and the face and neck region (24%).[4]

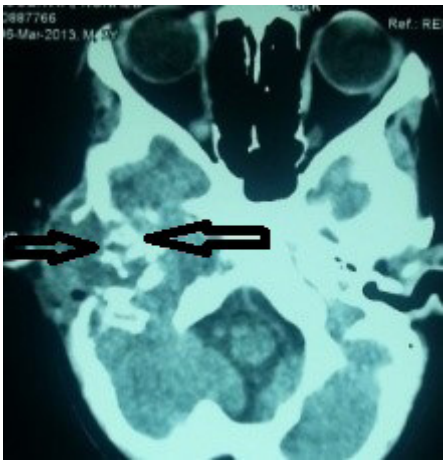
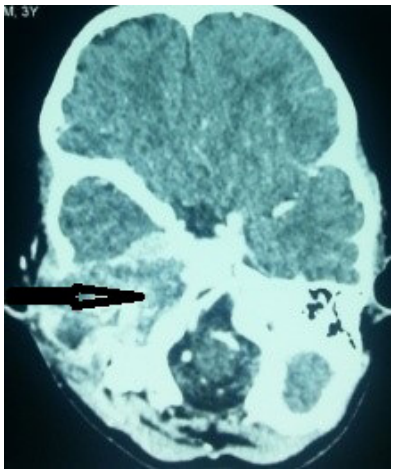


Figure 3. Pre-contrasted axial CT scans through the petrous part. Shows a soft tissue density mass in the petrous bone obliterating the external auditory canal.



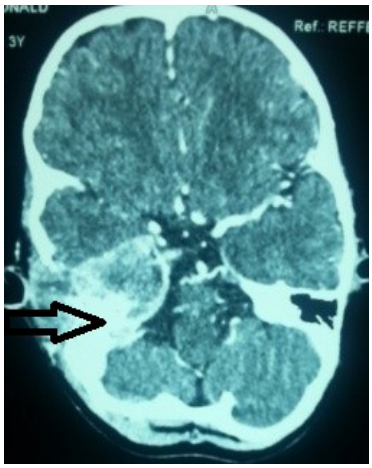


Figure 4a and 4b Axial sections shows heterogeneous enhancement of the mass. It is extending into the petrous apex.

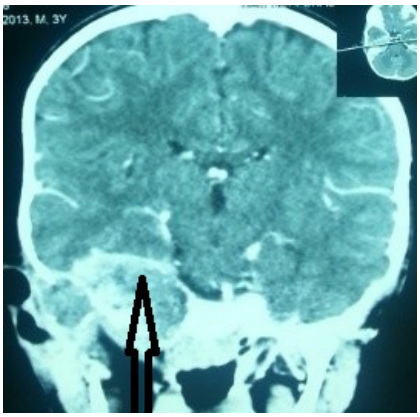


Figure 5. Coronal CT scans showing intra-cranial extension (extra-axial) of the mass with upward displacement of the overlying temporal lobe.

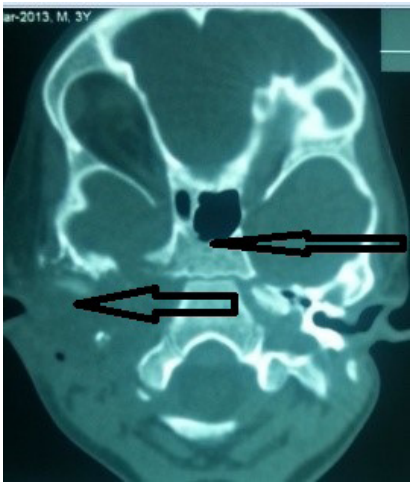


Figure 6. Bone window of axial CT scans shows destruction of the temporal bone (tympanic, petrous, and mastoid and posterior part of the squamous part). There is erosion of the clivus, tegmen tympani and sphenoid bone.

RMS that arise from the middle ear, may begin either in the muscles near the eustachian tube, in the proper middle ear, or from primitive pluripotential mesenchymal rests. At the time of diagnosis, there is usually widespread local invasion throughout the petrous bone and the actual site of origin is often obscure.[13] In two thirds of RMS cases arising from the middle ear there is already extensive bone erosion.[5,13] RMS of the head and neck with meningeal involvement carries the worst prognosis.[5] In our case there was meningeal involvement and the site of origin was not clear due to extensive destruction.

The main predisposing factor for the development of an RMS is a genetic mutation associated with 2:13 or 1:13 chromosomal translocation which produce PAX3-FKHR and PAX7-FKHR fusion products, respectively. These translocations result in altered expression and consequently contribute to its oncogenic characteristics. This is particularly true for alveolar RMS while in most embryonal rhabdomyosarcoma cases there is allelic loss at chromosome 11p15.[2]

Several pathological types of RMS have been described [1,8,9]:

1. Embryonal RMS (85%). This arises from the embryonic mesenchymal tissue in approximately 35% of all paediatric RMS of the head and neck.
2. Alveolar RMS (15%). Most often seen in older children characterized by faster growth than embryonic RMS and usually requires more intensive treatment.
3. Anaplastic RMS and undifferentiated sarcoma (pleomorphic RMS)

RMS usually presents with a triad of otitis media, bloody ear discharge and a polypoid mass in the external ear. Less commonly there is an external mass behind the ear. Occasionally, paralysis or paresis of the facial nerve occurs.[12] Progression is rapid with involvement of the petrous apex, internal auditory canal, and skull base leading to other craniopathies, estimated at approximately 30% of patients at the time of diagnosis.[10]

Middle ear inflammatory disease may be present at the same time, thus delaying the diagnosis and making total resection impossible in most cases. Chemotherapy and radiotherapy with or without tumour resection have resulted in improved prognosis.[12]

There are three commonly used clinical staging systems for RMS:[11]

1. IRS (Intergroup Rhabdomyosarcoma Study)
2. TNM, and
3. Clinical (surgico-pathologic).

The IRS classification includes four groups, based on whether the tumour can be resected:

i. Localized disease, tumour resected completely, regional lymph nodes not involved. Group I tumours have better prognosis

ii. Localized disease with microscopic residual disease or regional disease with or without microscopic residual disease.

iii. Incomplete resection with gross residual disease.

iv. Metastatic disease.

Groups iii and iv have the worst outlook and around 50% of RMS patients die following chemotherapy one year or so after histopathological diagnosis.[2, 6]

The diagnosis of RMS is usually confirmed by histopathology while other investigations such as MRI are used to evaluate the primary lesion and to rule out metastatic disease. CT Scan may be useful to determine bony erosion of the skull but the later (MRI) is superior in assessment of chest metastasis.

Ultrasound of neck in our patient showed cervical lymph nodes suggestive of metastasis.

Prognosis

Although the prognosis of this tumour used to be extremely poor (approximately 10% of patients survived five years), marked improvement in the survival rates has been reported over the past 30 years, particularly with the introduction of multi-modality therapy, in which surgery, multi-agent chemotherapy and radiotherapy have been combined.[11] Several factors may contribute to poor prognosis of the tumour such as old age, delayed diagnosis, and lack of understanding by the family as in our case.[14] Treatment, therefore, is by a multidisciplinary approach, consisting of surgical removal of the tumour followed by multi-agent chemotherapy with or without radiotherapy since RMS tends to metastasize to bone marrow.[12]

Conclusion

In paediatrics the presentation of RMS mimics complicated CSOM and hence may delay diagnosis. So, this rare tumour should always be suspected. A thorough evaluation and assessment is mandatory as it determines the management modalities mentioned to improve survival rates among patients.

The parents consented to publication of this case including the photographs (taken by Dr Justin Rubena Lumaya) and there is no conflict of interest in this case report.

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