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Case Report

Pathology Section

Embryonal Rhabdomyosarcoma: A Tale of Two Cases with Unusual Presentation

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ABSTRACT

Rhabdomyosarcoma (RMS) is a rare soft tissue tumour, accounting for 3% of all childhood cancers. RMS can arise virtually anywhere in the body, as it originates in embryonal mesenchyme. Embryonal Rhabdomyosarcoma (ERMS) and Alveolar Rhabdomyosarcoma (ARMS) are the most prevalent subtypes of RMS but arise through diverse biological mechanisms. RMS generally presents as an expanding mass that tends to be very painful and causes symptoms related to the compression of structures present nearby. Metastases of such tumours are common and can occur in bone, lungs and other organs leading to pain, difficulty with respiration, pleural effusion, anaemia, thrombocytopenia, and neutropenia. In the present report, two unusual presentations of ERMS have been reported. First case is of a 28-year-old adult male who presented in surgical Outpatient Department (OPD) with recurrent episodes of painful acute urinary retention since one year. Another case was of a six-year-old girl presenting with aural fullness and serosanginous discharge from right ear, ear pain and decreased hearing since two years. Since five year survival rate of such tumour is less than 30%, therefore, an awareness of the typical signs and symptoms, radiological features, histomorphological features in a case of paediatric and adult ERMS can help a pathologist to consider this tumour in the differential diagnoses, even at unusual sites.

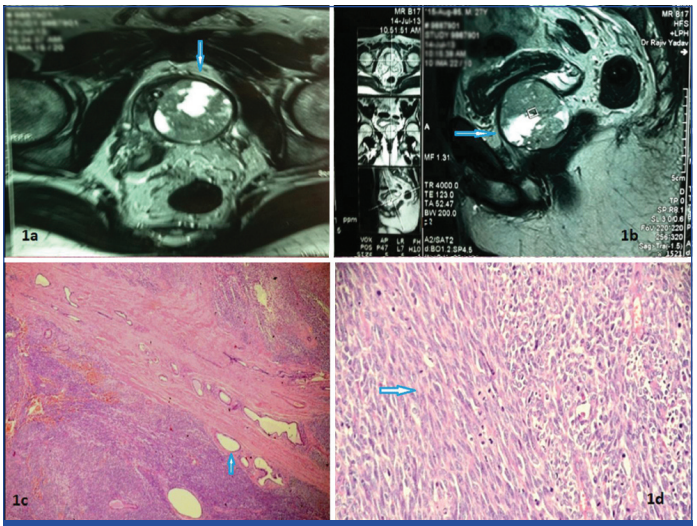
Keywords: Aural, Mastoid, Polyp, Prostate, Soft tissue tumour

CASE REPORT

Case 1

A 28-year-old male presented in the Department of Surgery with on and off episodes of painful micturition and inability to empty bladder completely since one year. There was no significant past medical or family history. Digital rectal examination revealed a smooth palpable cystic mass underlying the anterior rectal wall. Rectal mucosa was free of lesion. Computed Tomography (CT) urography revealed a large heterogeneously enhancing lesion measuring 5.5×5 cm in the prostate gland on left side causing displacement of urethra to right side [Table/Fig-1a]. Magnetic Resonance Imaging (MRI) prostate with spectroscopy revealed a large solid cystic mass in left side of prostate showing heterogenous enhancement, restricted diffusion, high choline and no periprostatic spread or significant lymph nodes [Table/Fig-1b]. Fine Needle Aspiration Cytology (FNAC) was performed at a peripheral lab and reported as a spindle cell neoplasm displaying a cellular smear composed of sheets and nests of spindle cells and few small round blue cells with scant cytoplasm along with frequent mitotic figures.

The patient open radical prostatectomy with vesicourethral anastamosis with bilateral pelvic lymph node dissection. Prostatectomy specimen measuring 5×4.5×4.5 cm was received. Cut surface showed a tumour measuring 4.5×4 cm involving almost the entire prostate. Bilateral seminal vesicles and vas deferens were free of tumour. On histopathology, tumour was arranged in interlacing fascicles interspersed by focal areas of necrosis. Tumour cells displayed spindle shaped nuclei, vesicular chromatin, inconspicuous nucleoli and scant amount of eosinophilic cytoplasm. Brisk mitotic activity was seen. Normal prostatic acini were noted in the peripheral area [Table/Fig-1c,d,2a]. Based on the histopathology, differential diagnosis of leiomyosarcoma was kept with a low suspicion of prostatic adenocarcinoma undergoing sarcomatous transformation. Hence, additional sections were taken to look for areas of carcinoma. Extensive sampling yielded no focus of adenocarcinoma. On Immunohistochemistry (IHC), tumour cells displayed diffuse positivity for vimentin, focal positivity for desmin and negativity for Smooth Muscle Actin (SMA). Owing to negative staining of SMA, possibility of leiomyosarcoma was excluded.

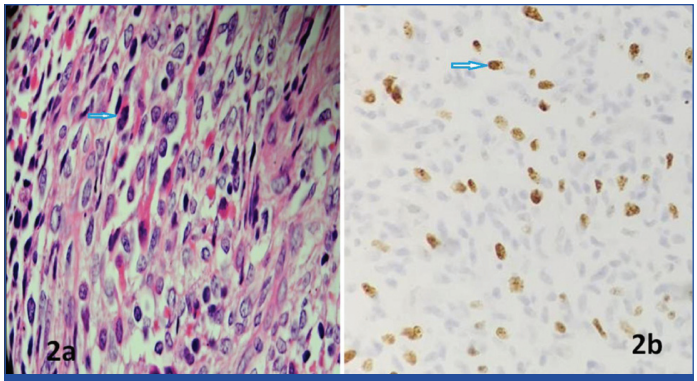


[Table/Fig-1]: a) CT urography showing a large heterogeneously enhancing lesion in the prostate gland on left side causing displacement of urethra to right side (arrow); b) MRI prostate with spectroscopy revealed a large solid cystic mass (arrow) in left side of prostate showing heterogenous enhancement, restricted diffusion, high choline and no periprostatic spread or significant lymph nodes; c) Histopathological Examination (HPE) of prostatic tissue showing tumour mass along with normal prostatic acini (arrow) seen in the periphery (40×, H&E); d) HPE of prostatic tissue showing tumour disposed in interlacing fascicles (arrow) (100×, H&E).

A closer inspection showed a small focus showing round to oval cells with abundant amount of eosinophilic cytoplasm along with occasional strap shaped eosinophilic rhabdomyoblasts. An extended panel including myogenin was applied which showed diffuse nuclear positivity [Table/Fig-2b]. Thus, owing to the predominance of spindle cells, focal areas of rhabdomyoblasts and IHC positivity for myogenin, the present case was reported as spindle cell variant of ERMS. Patient underwent radiotherapy, but was, lost to follow-up.

Case 2

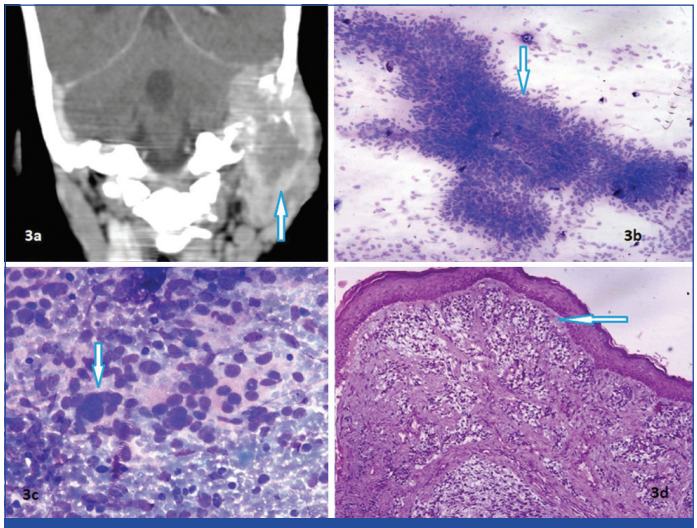
A six-year-old girl presented with blood tinged discharge from right ear, ear pain and decreased hearing for 15 days. She had aural



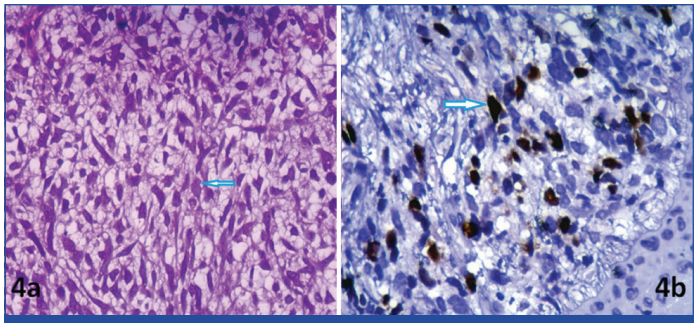
[Table/Fig-2]: a) HPE showing tumour cells displaying spindle shaped nuclei, coarse chromatin, inconspicuous nucleoli and moderate amount of eosinophilic cytoplasm. Occasional strap cells were noted (arrow) (400x, H&E); b) IHC showed diffuse nuclear positivity for myogenin (arrow).

fullness for past two years. On examination, a mastoid swelling measuring 4×4 cm was seen which was non tender, mobile with normal appearing overlying skin. Otological examination revealed an aural polyp measuring 2×2×1 cm protruding from the external auditory meatus. The CT scan showed an ill-defined heterogenous mass of size 6×4 cm with hypodense centre which was eroding the mastoid bone [Table/Fig-3a].

Fine Needle Aspiration Cytology (FNAC) was performed from the postauricular mastoid swelling which was highly cellular and revealed diffusely scattered medium sized round, oval to spindle shaped cells with hyperchromatic nuclei and scant amount of cytoplasm. Occasional rhabdomyoblasts were also noted [Table/Fig-3b,c]. Based on cytomorphology and clinical profile, a provisional diagnosis of small round blue cell tumour, most likely ERMS was made. The aural polyp was excised and mastoidectomy was done and excised tissue was sent for histopathological examination. Both the tissues showed similar histomorphology. The tumour was lined by startified squamous epithelium with a cellular cambium layer underneath it. Diffusely scattered tumour was noted in a background of loose, myxoid stroma. Tumour cells were round, oval to spindle in shape with hyperchromatic nuclei, inconspicuous nucleoli and scant amount of cytoplasm. Occasional strap cells were also noted [Table/Fig-3d,4a]. IHC showed diffuse cytoplasmic positivity for vimentin, nuclear positivity for myogenin [Table/Fig-4b]. The diagnosis of ERMS was confirmed. Patient was lost to follow-up.



[Table/Fig-3]: a) CT scan showing a mass eroding the mastoid bone (arrow); b) FNAC of postauricular mastoid swelling showing dyscohesive cluster and singly scattered medium sized round, oval to spindle shaped cells with hyperchromatic nuclei and scant amount of cytoplasm (arrow). (100×, May Grunwald-Giemsa (MGG)); c) FNAC of postauricular mastoid swelling revealing medium sized round, oval to spindle shaped cells with coarse chromatin and scant amount of cytoplasm, few multinucleated giant cells noted (arrow). (400×, MGG); d) HPE of postauricular mastoid swelling showing stratified squamous epithelium with a cellular cambium layer (arrow) underneath it. Diffusely scattered tumour was noted in a background of loose, myxoid stroma (100×, H&E).



[Table/Fig-4]: a) HPE showing round, oval to spindle shaped cells displaying coarse nuclear chromatin, inconspicuous nucleoli and scant amount of cytoplasm (arrow).(400×, H&E); b) IHC showed nuclear positivity for myogenin (arrow).

DISCUSSION

The RMS can develop in almost any part of the body, most common site being genitourinary organs accounting for 29% of the cases. The orbit is the most usual location for paediatric RMS, but it can occur in the oral cavity, pharynx, face, and neck in descending order of incidences [1]. Involvement of the ear and temporal bone is uncommon [2]. RMS has a broad range of reported age range between 17-68 years and only few cases are reported in adults [2,3]. Embryonal, botryoid, alveolar, pleomorphic, spindle cell and anaplastic variants are the pathologic subtypes of RMS and ERMS and comprises of about 60-70% of RMS [4]. Though ERMS is generally regarded as a neoplasm occurring primarily during childhood, but there are few reports in the literature that occurred in adults [5]. Prostate ERMS is a common tumour in infants and children, with a median occurrence age of 5 years [6]. A 15-20% of all RMSs arise from the genitourinary tract. It has the proclivity to involve the hollow organs like bladder, prostate, paratesticular region, uterus and vagina and it is the 3rd most common extracranial solid tumour of childhood after nephroblastoma and neuroblastoma [7]. The common variants of sarcomas affecting the prostate include leiomyosarcoma, RMS, malignant fibrous histiocytoma and unclassified sarcoma. Among all these, RMS is the most common histological subtype that accounts for approximately 40% of prostate sarcomas in children. Leimyosarcoma accounts for nearly 25% cases and mostly occurs in the elderly [8]. When an adult male presents with lower urinary tract obstruction and markedly enlarged prostate with normal Prostate-Specific Antigen (PSA) levels, prostate sarcoma is often suspected [9].

Spindle cell lesions of prostate poses a great diagnostic challenge as it encompass a broad range of benign and malignant tumours originating from the prostate epithelium or stroma, such as sclerosing adenosis, sarcomatoid carcinoma, Stromal Tumours of Uncertain Malignant Potential (STUMP), stromal sarcoma, Solitary Fibrous Tumour (SFT), leiomyosarcoma, Inflammatory Myofibroblastic Tumour (IMT), Gastrointestinal Stromal Tumour (GIST), fibrosarcoma and Malignant Peripheral Nerve Sheath Tumour (MPNST) [10]. Identification of benign or malignant nature of tumour cells along with admixture of benign or malignant glands and the presence of heterologous elements are the few useful features that aids in distinguishing these lesions from one another. Admixture of prostate glands with the spindle cell lesion poses a more specific differential diagnosis as in the present case like some patterns of STUMP, stromal sarcomas (malignant phyllodes pattern), sarcomatoid carcinoma and sclerosing adenosis. In sclerosing adenosis, STUMPs and stromal sarcoma, the glandular component is typically benign, whereas sarcomatoid carcinoma shows admixed adenocarcinoma component. Rest of the lesions like IMTs, leiomyosarcomas, GISTs and SFTs generally grow in an expansile pattern without any admixed glandular element. In such pure spindle cell lesions, cytological features and growth pattern are two important features in diagnosis of these entities [9]. The recognition of rhabdomyoblast is the key to the diagnosis of RMS. However, in more primitive tumours, IHC such as myogenin, myo-D1 and desmin can help to confirm diagnosis of RMS [10].

Clinical features of ERMS include rapidly progressing obstructive urinary symptoms, enlargement of prostate on digital rectal examination, infrequent suprapubic mass, involvement of nearby lymph nodes, difficulty in breathing due to lung metastasis, osteoclastic bone metastasis and normal serological markers like PSA levels and prostatic acid phosphates [11,12]. Botryoide type of ERMS is the usual presentation of urogenital RMS in children and they respond well to radiation and chemotherapy [13]. In contrast, adults usually present with non embryonal subtypes which tend to be widely disseminated. The results are poor and despite good initial responses to chemotherapy, they eventually die of their disease [14]. The lack of awareness of this entity in adults, delay in diagnosis and more aggressive behaviour of this malignancy are some of the reasons [3]. Localised presentation and favourable prognosis of index case was due to the histological subtype. Cases with spindle cell morphology of ERMS are extremely rare and till date, only four similar cases have been reported in literature, index case is the fifth such case. Schildhaus HU et al., published a case of 25-year-old male with problem of micturition which was reported as spindle cell variant of ERMS [15].

Rhabdomyosarcoma (RMS) in the head and neck can occur in the orbit, pterygopalatine fossa, parapharyngeal space, nasopharynx but very rarely in the middle ear and mastoid. RMS of mastoid is a deadly neoplasm that occurs almost exclusively in paediatric age group. Hence, the misdiagnosis as aural polyp is generally made. Therefore, such tumour presents as advanced disease at the time of diagnosis [16]. Clinical features of such tumours include purulent, blood tinged ear discharge, hearing impairment, earache, aural polyp. Advanced cases may present with neurologic symptoms. As these features are non specific, they can lead to a delayed diagnosis. Therefore, most of these patients are initially managed on antibiotics and only when this treatment fails, other diagnosis is suspected [17].

In 1966, Potter reported a 3-year-old male with bilateral otitis media and polypoidal external auditory canal mass that turned out to be RMS [18]. Metastases of such tumours commonly spread to the lungs, liver, bones and extremities, and are present in approximately 30% of cases [19]. The main treatment is often chemo-radiotherapy. Parameningeal tumours, especially of the middle ear and mastoid have a poor prognosis because of the involvement of the brain [20].

CONCLUSION(S)

The possibility of prostatic RMS should be kept in mind when a young male presents with prostate mass, lower urinary tract symptom and normal PSA. Likewise the diagnosis of RMS should be sought for in a child presenting with mastoiditis and not responding to the routine therapy. A high index of suspicion is needed to avert a grave prognosis. Timely detection and early diagnosis are the key to successful management of these sarcomas at rare sites.

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