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A Comprehensive Case Report on Embryonal Rhabdomyosarcoma

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ABSTRACT

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in the pediatric and adolescent population. While they can develop anywhere in the body, the genitourinary tract is the second most common primary location for an RMS to develop. Here we report a case of 2 year old male patient who have presented with the complaints of perianal rash and difficulty in voiding for 2 weeks. According to the histopathological and immunohistochemical report, it was diagnosed as an embryonic variety of rhabdomyosarcoma. The child was categorised as being in the group lll, stage lll, intermediate risk category of embryonal RMS. Furthermore, we discuss about the clinical, radiological, immunohistochemical findings, and management of the aforementioned RMS case.

Keywords: Rhabdomyosarcoma, cystoprostatectomy, immunohistochemistry, chemotherapy

INTRODUCTION

Rhabdomyosarcomas (RMS) are malignant soft tissue tumors of primitive mesenchymal cells that undergo partial rhabdomyoblast differentiation and is the second most common sarcoma in children after osteosarcoma. RMS accounts for 4–8% of all malignant illness cases in children under the age of 15. These tumors have a bimodal distribution and frequently manifest in patients between the ages of 10 and 18 as well as younger than 5 years old. The genitourinary tract, retroperitoneum, oral and craniofacial region, and extremities are the most often involved sites of RMSs [1,2]. There are 350 new cases of rhabdomyosarcoma (RMS) reported annually, making it the most prevalent soft tissue sarcoma in the paediatric and teenage population. Rhabdomyosarcomas (RMS) can show up in various body parts, but they often appear in the trunk, arms, legs, or head/neck. About 18–22% occur in the genitourinary tract, which includes organs like the bladder, kidney, and reproductive areas, making it the second most common location for RMS [3].

RMSs in the GU tract is often categorised as non-bladder/prostate RMSs or bladder/prostate (BP-RMSs). Male non-bladder/prostate RMSs are paratesticular, whereas female non-bladder/prostate (FGU-RMS) primaries originate from the vagina, vulva, and/or uterus [4].

It might be challenging to determine the exact origin of BP-RMSs and other pelvic RMS tumors because of the close proximity of the affected organs [5]. Hematuria, dysuria, and urinary blockage are examples of genitourinary symptoms that can be brought on by BP-RMSs. In line with RMSs originating from different places, BP-RMSs can likewise elicit symptoms secondary to mass impact (e.g., constipation) or be asymptomatic [6].

CASE REPORT

A 2-year-old male patient, born full term via spontaneous vaginal delivery, with no significant past medical history, presented to the emergency center with chief complaints of perianal rash, which was thought to be diaper rash, and had difficulty voiding for two weeks. Consequently, pressure was applied to void since he could not void. Due to bladder obstruction, urinary output decreased, and a catheter was inserted for 10 days and changed for two times. Blood was found in urine, but there was no history suggestive of fleshy pieces in urine.

The patient weighed 12 kg and showed typical age-appropriate growth and development. There was no prior history of fever, changes in bowel habits, weight loss, or decreased appetite. Upon examination, there was no pallor, cyanosis, clubbing, or edema, and the patient was vitally stable and afebrile. A hard palpable mass in the suprapubic region was discovered during a physical examination of the abdomen and genitalia. A hard mass was felt anterior to the rectum and no mass was felt posterior to the region, according to the rectal examination.

Urinalysis revealed hematuria of >3 RBC/hpf with no signs of infection. Hematological and biochemical investigations were normal. A USG of the whole abdomen revealed a 68×59×47mm-sized heteroechoic lesion posterior to the urinary bladder, likely in the presacral region, and suggested further evaluation. A CECT whole abdomen was recommended; this confirmed approximately same sized ill-defined irregular outline solid focal mass lesion seen in the pelvic cavity at presacral space. And this showed that the prostatic urethra is encased by the lesion and that there is no intraspinal or extra-pelvic extension. The heterogeneously enhancing mass lesion in the pelvic cavity shows an ill-defined fat plane with the posterior wall of the urinary bladder and rectum, with involvement of the prostatic urethra. A chest x-ray showed that both lungs don't have any parenchymal lesions and have an intact bony thorax. The liver is normal in size with normal echotexture and doesn't have any focal lesions.

The patient underwent a cystoscopy, and this revealed normal bladder mucosa and anterior urethra. Due to the compression of mass from behind the posterior urethra, it is elongated very steeply. so unable to negotiate scope across the posterior urethra. Hence, it was decided to proceed with an open biopsy after discussion. A biopsy is taken from the mass posterior to the bladder. A vesicostomy was made by a lower vertical incision, and an 8-F urethral catheter was placed. The patient was treated with antibiotics and analgesics. (inj. cefuroxime 500mg BD and inj. paracetamol 180 mg TDS)

Histopathological examination showed the presence of elongated cigar-shaped cells with pleomorphic nuclei and skeletal muscle-like cross-striations in the cytoplasm, characteristic of rhabdomayoblasts, along with an abundant population of small, pleomorphic round cells, numerous giant cells, and spindle cells, suggestive of RMS. Immunohistochemical marker tests were performed to better clarify the lesion, and the report showed tumor cells were positive for desmin and vimentin. According to the histopathological and immunohistochemical report, it was diagnosed as an embryonic variety of rhabdomyosarcoma. The child was categorised as being in the group lll, stage lll, intermediate risk category of embryonal RMS.

The patient was scheduled for surgical excision of the tumor, followed by adjuvant chemotherapy. The child was treated as per the children's oncology group (COG) protocol and immediately started on the VAC chemotherapy regimen. He underwent 14 cycles of vincristine, dactinomycin, and cyclophosphamide (VAC regimen). He received one dose of granulocyte-stimulating factor after each cycle in view of significant neutropenia. Prior to individual chemotherapy, medications such as antihistamines and antiemetics must be given to cope with untoward symptoms. At the end of the therapy, the CT scan was stable and showed no active disease. In view of hypertension (115/97 mmHg) after the last cycle of chemotherapy, the child was prescribed Tab. Nifedipine 2.5 mg TDS.

After all preoperative investigations, informed consent, and pre-anaesthetic evaluation, the child underwent excision of rhabdomyosarcoma with radical cystoprostatectomy and bilateral end ureterostomies (Wallace technique). Ureterostomies matured at umbilicus using 5-0 vicryl and both ureters stented with 3Fr 8 cm (R); 10 cm (L) DJ stents and connected to colobag. After proper hemostasis, the wound was closed, and his recovery was uneventful.

The intraoperative and postoperative periods were uneventful. Post-operatively, the child was treated with antibiotics and analgesics. The child was clinically and hemodynamically stable and ultimately discharged.

DISCUSSION

Rhabdomyosarcoma (RMS) is a primitive paediatric malignant soft tissue sarcoma of the skeletal muscle phenotype that originates from a primitive mesenchymal cell. Most of the aetiology and risk factors are still unclear. In children, the RMS is an aggressive tumor that grows quickly. RMS is more common in children between the ages of 1-4 and 10–14 [7]. The impact on men is 1.5 times greater than that on women. our patient was a 2 -year-old boy and the tumor was located in the posterior wall of the urinary bladder and rectum, with involvement of the prostatic urethra.

A careful histological and immune-histochemical diagnosis are necessary to exclude differential diagnostic cases resembling RMS, such as Ewing's sarcoma, spindle cell sarcoma and osteosarcoma [8]. Several immunohistochemical markers have been applied for diagnosis of RMS, among those, desmin and vimentin were considered as two prominent and useful markers for RMS, and help to differentiate from the other soft tissue tumors [9]. The tumor cells of the current case were tested positive for desmin and vimentin, Different treatment modalities like surgery, radiotherapy and different regimens of chemotherapy are used for the treatment of rhabdomyosarcoma. The treatments are based on the stage and clinical presentations of the tumor. Several prognostic indicators have been identified, which include age, tumor location, and histologic type [10].

According to histopathological & immunohistochemistry report it was diagnosed as embryonal variety of Rhabdomyosarcoma. The management of RMS includes multimodal therapy, where chemotherapy plays an important role and often associated with radiotherapy depending on the severity of the disease. Two main chemotherapy regimens used in the treatment of rhabdomyosarcoma include (1) the VAC regimen, which consists of vincristine, actinomycin D, and cyclophosphamide, and (2) the IVA regimen, which consists of ifosfamide, vincristine, and actinomycin D. These drugs are administered in up to 14 cycles, depending on disease stratification [11].

Initial treatment of rhabdomyosarcoma involves complete gross surgical excision. The goal of surgical treatment is to remove the tumor without causing a major functional or cosmetic defect. In our presented case, we have treated the case by surgical excision of tumor followed by adjuvant chemotherapy with Vincristine(1.5 mg/m2) dactinomycin (0.045mg/kg),

and cyclophosphamide(2200mg/m2) (VAC regimen) up to 14 cycles(40 weeks). To reduce the tumor size the patient underwent chemotherapy and secondly with cystoprostatectomy and bilateral end ureterostomies which brings the two ureters to the surface of the abdomen, one on each side.

This case highlights several important characteristics for young children with GU RMS. An embryonal RMS (ERMS) generally carries a favorable prognosis and consists of oval-spindle-shaped cells with morphologic features resembling fetal skeletal muscles. It accounts for 80% of all GU RMSs.

Nonetheless, a number of studies show that being younger than five years old when diagnosed significantly worsens the prognosis. Furthermore, while the patient is still very young, radiation treatment may have terrible side effects include intestinal radiation and stunted growth. Local management, including surgical resection where required, is crucial to the successful treatment of children less than five years at the time of diagnosis.

CONCLUSION

Although in general, the incidence of rhabdomyosarcoma is more common in children between the ages of 1-4 and 10–14. Its propensity to invade the genito-urinary tract makes it a troublesome clinical entity to deal with. A careful histological and immune-histochemical diagnosis are necessary to exclude differential diagnostic cases resembling RMS, such as Ewing's sarcoma, spindle cell sarcoma and osteosarcoma. Several immunohistochemical markers have been applied for diagnosis of RMS, among those, desmin and vimentin were considered as two prominent and useful markers for RMS. So proper chemotherapeutic regimen, preoperative evaluation, preoperative assessment with complete resection, and long term follow-up is the mainstay of management of this condition.

Declaration by Authors

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