Case Report

Perianal Rhabdomyosarcoma Presenting Post Surgery for Hirschsprung's Disease: A Rare Presentation

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Keywords: *Cytopathology, Hirschsprung's, Perineal, Rhabdomyosarcoma*

ABSTRACT

Perianal region is an uncommon site for Rhabdomyosarcoma. Early diagnosis is essential in these cases, as 25% of patients present with metastasis at the time of diagnosis. FNAC is simple and useful procedure for making an early diagnosis and helping clinicians for prompt intervention. Previously, a single case had been reported of rhabdomyosarcoma occuring 2 years after a surgery for hamartoma at the same site. Herein, we report a case of perianal rhabdomyosarcoma which occurred 5 years after a pull through procedure done for Hirschsprungs disease and this rare association has not been reported yet in literature.

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Introduction

Perineal or perianal rhabdomyosarcoma are extremely rare and occur in <2% of primary cases. Overall, rhabdomyosarcoma is the most common soft tissue sarcoma of childhood with an incidence of 4-7 per million in the age group of 15 years or younger. The common sites of involvement are head and neck, genitourinary tract, extremities with perianal region being an uncommon location. They usually have regional lymph node metastasis at the presentation and comparatively poor prognosis. Many a time misdiagnosed as perirectal or gluteal abscess delaying the diagnosis and treatment. Previously, a single case had been reported of rhabdomyosarcoma occurring 2 years after a surgery for hamartoma at the same site [1]. Herein, we report a case of perianal rhabdomyosarcoma which occurred 5 years after a pull through procedure done for Hirschsprungs disease. No such association has been reported till date which makes the present case to be of its first kind to be published.

Case Report

A seven year old female child presented with an irregular, polypoidal perianal mass of 2 months duration. The patient also had a history of Hirschsprungs disease at 1.5 years of age, which was diagnosed on histopathological examination. She had undergone a pull through procedure for the correction of disease 5 years back. Following surgery, she did not have any problem. But recently, a rapidly growing mass was detected by the patient's parents in last 2 months. On examination, the mass was lobulated, non-tender mass measuring 6X4.5 cms with normal appearing overlying skin (Fig1A). The mass was external to the anal opening and no abnormality was detected on per-rectal examination. Clinically the provisional diagnosis was anal papilloma or melanoma.

Fine needle aspiration cytology examination done revealed small round cells with scant fragile cytoplasm, round nucleus and prominent nucleoli, suggestive of small blue round cell tumor (?) rhabdomyosarcoma (Fig 1B). Further to confirm the diagnosis made on cytopathological examination, a cell block was made. Smears showed round cells arranged in nests and sheets with scant cytoplasm, with prominent nucleoli and many mitotic figures (Fig 2A & B). Strap cells were also noted (Inset 2B).

Immunohistochemical studies confirmed the diagnosis of rhabdomyosarcoma with positive expression of Vimentin, Desmin and Myogenin (Fig 3A-C). However, Cytokeratin, LCA, NSE were negative (Fig 3D-F).

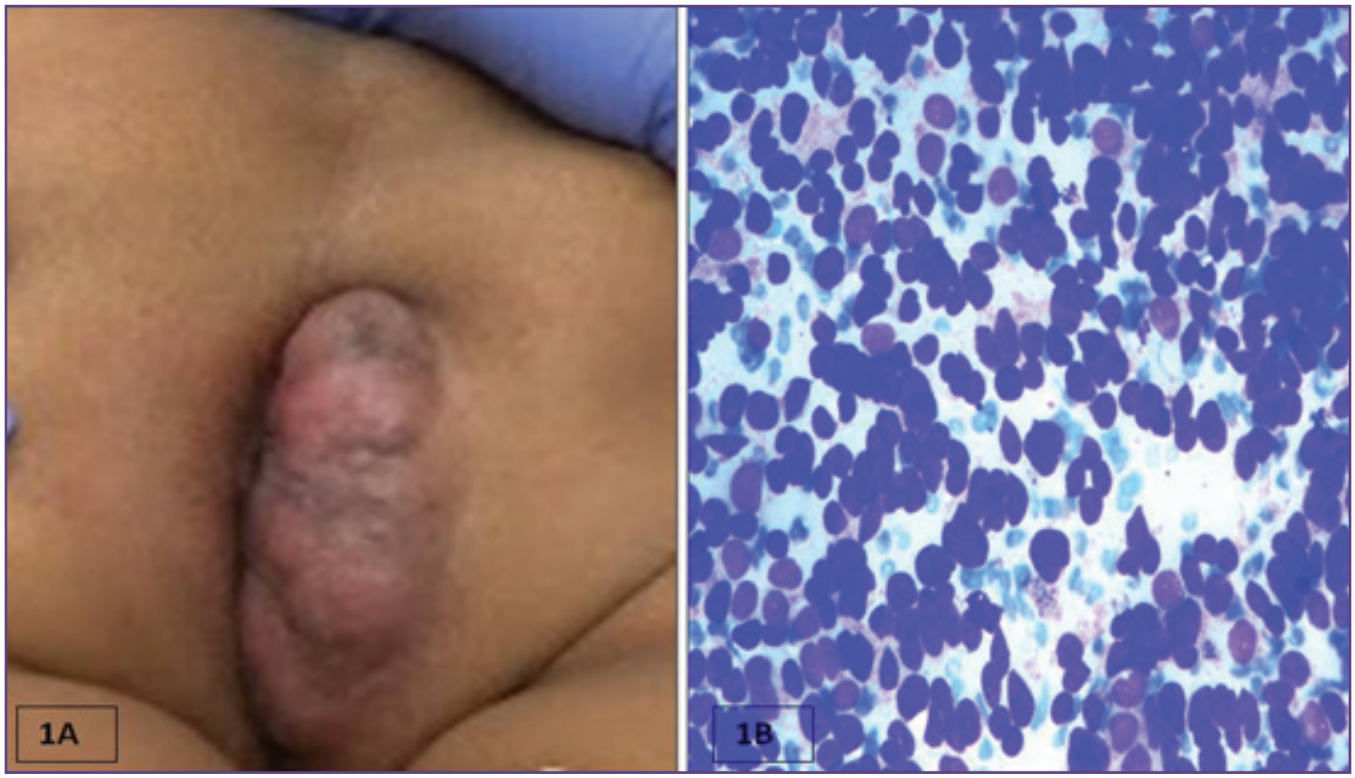

Fig. 1A: Gross picture of the lobulated tumor in perianal area. Fig 1 B: Smears showing small round cells with scant fragile cytoplasm, round nucleus and prominent nucleolus (Giemsa, X 400).

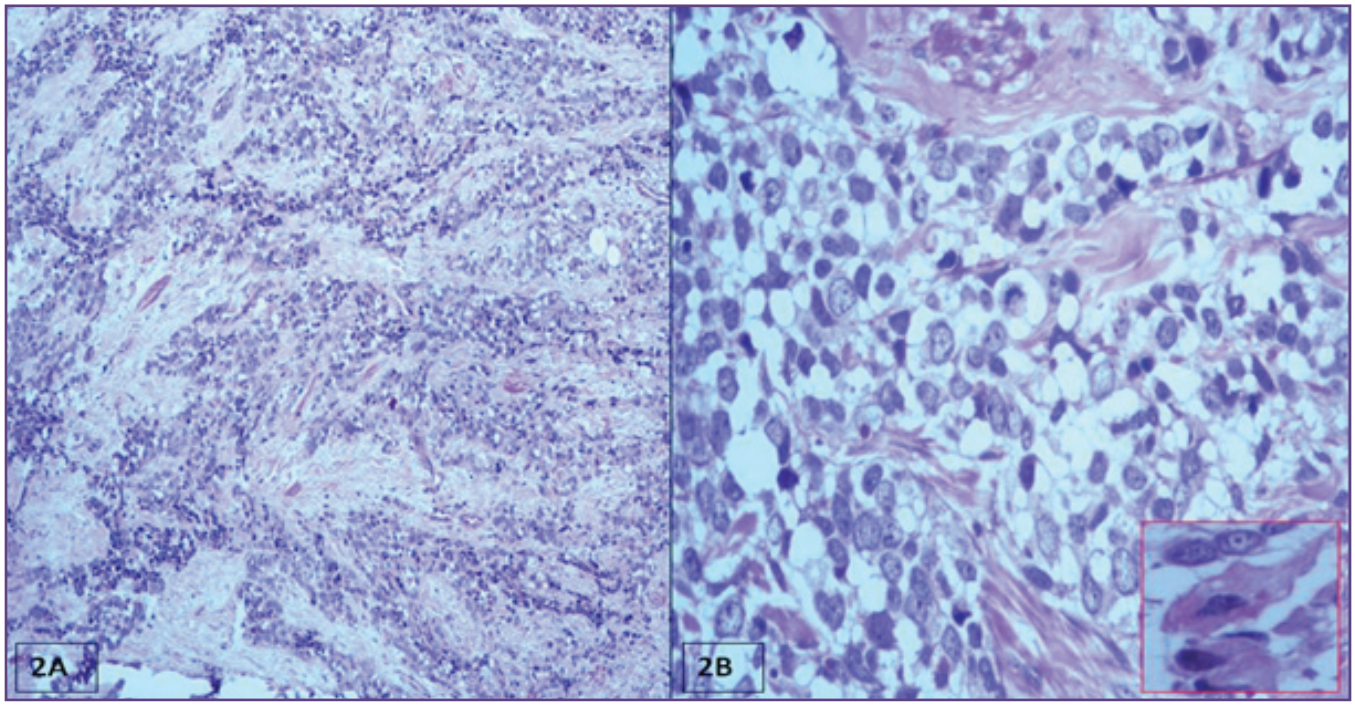

Fig. 2A & 2B: Sections showing round cells arranged in nests and sheets with scant cytoplasm , with prominent nucleolus and many mitotic figures. Inset (2 B) shows a strap cell. (2 A: H&E, X100; 2 B: H&E, X400).

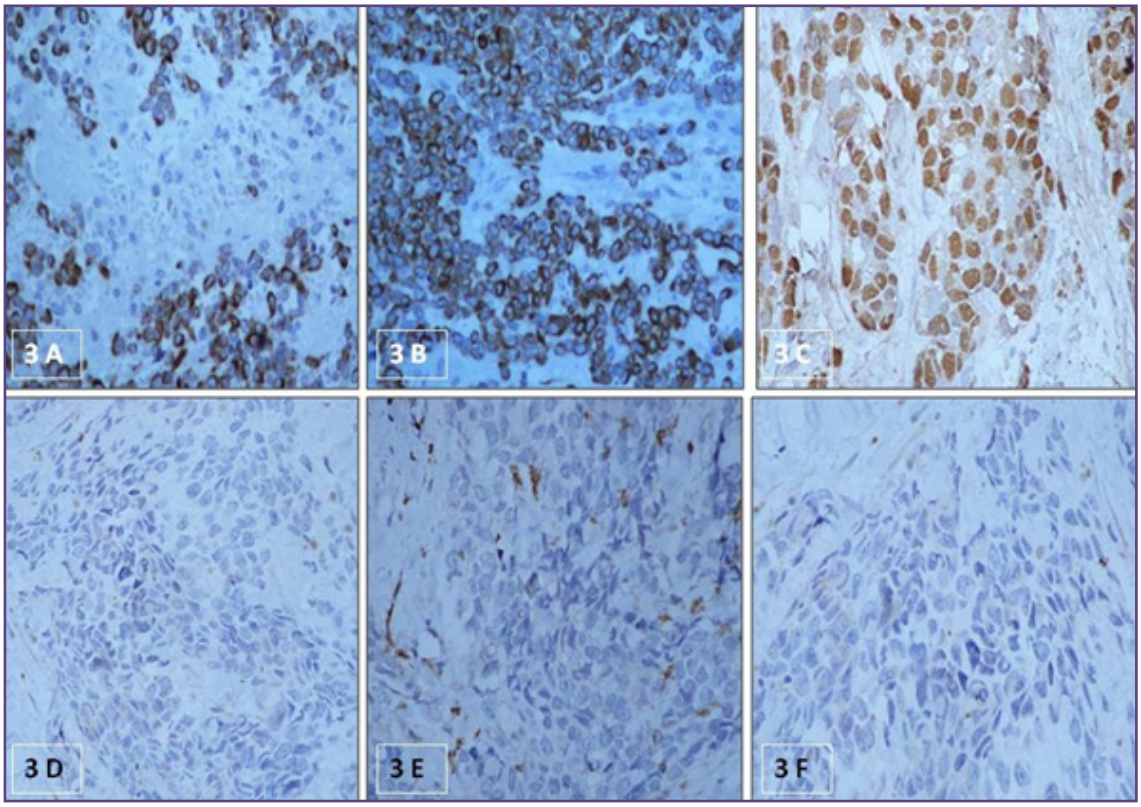

Fig. 3-C: Tumor cells showing immunoreactivity for Vimentin (3A), Desmin (3B) & Myogenin (3C), respectively. [X400]. Fig 3D-F: Photomicrograph showing negative expression of CK (3D), LCA (3E) & NSE (3F), respectively. [X400]

Considering the clinical, cytological, histopathological and immunohistochemical aspects, finally a diagnosis of Perianal Rhabdomyosarcoma (Embryonal type) occurring after pull through procedure done for Hirschsprungs disease was made. The patient was advised to undergo extensive surgical resection along with chemotherapy and was under follow up for only two months after which she was lost to follow up.

Discussion

Rhabdomyosarcoma is a malignant tumor of striated muscle origin and is the most common soft tissue sarcoma of childhood with an incidence of 4-7 per million children in the age group of fifteen years or younger [2,3]. The common sites of involvement are head & neck, genitourinary tract, extremities. Less commonly trunk, intrathoracic region, perineal/ perianal region, biliary tract are also involved. Perianal rhabdomyosarcoma is rare condition and rare cases of anorectal rhabdomyosarcoma have been reported in medical literature [4]. It is interesting to note that when we consider the lesions of somatic origin, leiomyoma or leiomyosarcoma are the common tumors and originating in lower portion of genitourinary system, rarely in anorectum. Tumors encountered in early period of life are usually of epithelial origin. Usually the lesions of somatic origin are rare including tumors of colon and rectum in children. Hence, considering its rarity in children, it becomes difficult to timely diagnose these lesions from clinical aspect.

The important misleading points are variation in growth characteristics, gross and histopathological findings. They may occur as apparently well-defined, encapsulated, perineal mass; easily getting confused with condyloma acuminata, fibromas or fibromyosarcoma. But the most important crux to be remembered is that the highly malignant behavior of these tumors is the key to rational therapy [5-6]. Similarly, gross features are important to be noted as the gross consistency in rhabdomyosarcoma varies in proportion to their collagen content. They are not as firm as fibroma or fibromyosarcoma. Sometimes they may present with reddish discoloration caused by hemorrhage or necrosis. There are cases in literature where perianal rhabdomyosarcoma presented with abscess [7].

The histopathological findings in rhabdomyosarcoma in children is different from that seen in adults. In children, the lesion appears to originate from primitive mesenchymal cells with marked cellular distortion and myxomatous elements. They grow very rapidly in infants and children giving a clue to that they are of embryonic origin arising from immature myeloblasts rather than mature striated muscle as seen in adults. The present case is the first of its kind to be reported as because association of perianal rhabdomyosarcoma occurring after pull through procedure for Hirschsprungs disease has never been reported in literature till date. However, there has been a report of perianal RMS after surgery for hamartoma at same site. Previously a single case had been reported of rhabdomyosarcoma occurring 2 years after a surgery for hamartoma at the same site [1]. Our case was after pull through surgery for Hirschprung's disease. No case was reported in the past relating the two.

It is important to differentiate this entity from other small blue round cell tumor which is noted in children. Hence, role of immunohistochemistry is of utmost importance which helps in excluding other entities. A proper approach using correct immunohistochemical markers help in specifically diagnosing the exact type of malignant process involved.

Perianal RMS are high-risk tumors. According to the Intergroup RMS Study Group (IRSG) review of 71 children with perineal or anal RMS from 1972 through to 1997, the prognosis is poor, with a 5-year failure-free survival rate of only 45% and overall survival (OS) rate of 49% Multimodal treatment should be initiated without delay [8]. A perianal site is unusual and is associated with high risk and a low cure rate [9-10]. The treatment of choice for RMS combines intensive chemotherapy, high-dose radiotherapy and complete surgical excision, but there is no established treatment strategy for RMS of the perineum or anus, in particularly in the Intergroup Rhabdomyosarcoma Staging (IRS) reports, as these locations are rare. The few cases of perianal RMS that have been reported have been associated with frequent sphincter disorders or anal ulcerations [11-12]. Wide first-line curative surgery is possible but causes loss of sphincter function. Multimodal treatment may preserve sphincter function and achieve remission without major complications, and should not be withheld because of young age.

Conclusion

Perianal region is a rare location for rhabdomyosarcoma and it can mimic some benign condition clinically as in the present case. Early diagnosis is essential in case of rhabdomyosarcoma, as 25% patients present with metastasis at the time of diagnosis. Fine needle aspiration cytology, core needle biopsy and immunohistochemistry should be used as a diagnostic tool for perianal mass lesion. The present case is unique because of its occurrence after five years of pull-through surgery for Hirschsprungs disease and is the first case to be published in literature.

Acknowledgements
None

Funding
None

Competing Interests
None declared.

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