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Congenital embryonal rhabdomyosarcoma; multiple lesions



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ABSTRACT

INTRODUCTION: Congenital or neonatal rhabdomyosarcoma (RMS) is a rare soft tissue tumor with the most common sites of origin in genitourinary tract, head, and neck regions and extremities are less commonly involved.

PRESENTATION OF CASE: In this paper, a case of embryonal RMS with skin lesions, lymph nodes metastasis, and bone marrow metastasis is reported for a 1-month old female patient.

DISCUSSION: This study presents how within 8-months of chemotherapy, the lesions got subsided and the patient became disease free.

CONCLUSION: Multiple congenital rhabdomyosarcoma of neonate is a rare finding that should be considered as differential diagnosis of lymphoma and neurofibroma.

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1. Introduction

Rhabdomyosarcoma (RMS), which is a neoplasm of soft tissue, is seen both in adults and children; however, its occurrence in infants is very rare [1]. RMS is seen mostly in head and neck regions, genitourinary system and extremities. The most common sites of metastatic involvement are the soft tissues, serosal surfaces, lung, bone marrow, and lymph nodes; therefore, it can be mistaken for acute leukemia and lymphoma [2]. To the best of our knowledge, less than 10 cases of congenital rhabdomyosarcoma have been reported up to this time. We present a case of congenital embryonal rhabdomyosarcoma with unusual clinical features of multiple skin nodules and bone marrow metastases.

2. Case report

A 6-month-old female, first child of a family, was presented to our department with irritability and skin lesions which appeared since she was 1-month-old. Her parents noticed these skin lesions as dark-purple swellings in different sizes, first at left extremity and short after throughout her body since she was 1-month-old (Fig. 1). Some of these lesions were waxed and waned over time, but didn't have been improved. Lab findings revealed high ESR: 88 and LDH: 8932 and low Hemoglobin: 7.9 g/dl levels. Blood glucose, liver function tests, electrolytes were all in normal limits. Vinyllylmandelic Acid (VMA) and homovanillic acid (HVA) tests

for neuroblastoma were unremarkable. CT scanning of brain, thoracic, abdomen and pelvic organs revealed multiple subcutaneous nodules in skull, chest wall, and abdominopelvic skin, some with necrotic centers and the largest measured at 39*50 mm. Multiple lymphadenopathies were noted in supraclavicular, both axilla, both iliac chain, inguinal and right paraaortic (27*15 mm). There weren't any significant organic abnormalities (fig2). Skin biopsy was performed; histopathologic studies revealed proliferation of small round dispersed cells, composed of primitive mesenchymal cells in various stages of myogenesis with scant to moderate amount of deeply eosinophilic cytoplasm with occasionally cross striations, eccentric, small oval nuclei, and inconspicuous nucleoli (Fig. 3). Immunohistochemical staining revealed strong positivity for desmin and myoglobin (Fig. 4); while, it was negative for CD1a, LCA, CD68, CD99, Synaptophysin, Chromogranin and WT1. IHC staining for S100 showed unspecific staining in tumor cell.

As the patient developed pancytopenia, she underwent bone marrow aspiration, which showed monotonous infiltration of large tadpole-like cells in bone marrow with abundant bluish cytoplasm (fig5). On the basis of histopathology, diagnosis of embryonal rhabdomyosarcoma was made. Due to the observation of multiple lesions, the patient underwent chemotherapy with Vincristine, Actinomycin, Cyclophosphamide, and Dexamethasone, and remained disease free for 8 months after diagnosis.

3. Discussion

Rhabdomyosarcoma is a soft tissue tumor which accounts for 4–8% of all soft tissue tumors in children mostly occurring between the ages of 3 and 12 years [1,8]; however, its occurrence in infancy is extremely rare [1,2,5,6]. Males are affected more often, but there is not gender difference in recent reported neonatal cases [4,7,8].

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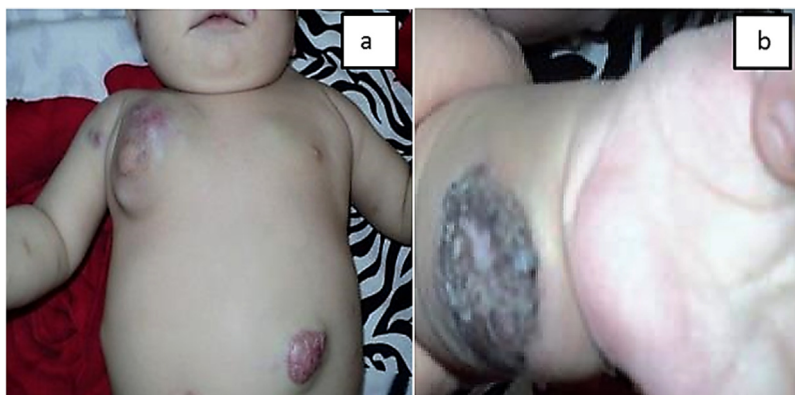


Fig. 1. a,b; Subcutaneous nodules of rhabdomyosarcoma in a 6 month old female.



Fig. 2. Magnetic resonance imaging (MRI) findings show large subcutaneous nodules with multiple lymphadenopathies, without organic abnormalities.

RMS is classified into three types of embryonal, alveolar and pleomorphic, as well as several subtypes and possibly minor categories [1–9]. Embryonal type variants and related tumors include botryoid, pleomorphic, clear cell, rhabdoid, spindle cell and sclerosing types [10–12].

Embryonal rhabdomyosarcoma is the most common type in infancy and children among above-mentioned three types [4,7]. This tumor type arises from undifferentiated mesoderm and is

common in the head and neck region (particularly the orbit, nasopharynx, middle ear, and oral cavity), retroperitoneum, bile ducts, and urogenital tract [7,13]. Less commonly it may occur in the extremities and skin. Among these sites, survival rate is less in those with affected extremities [14]. Clinically, its differential diagnoses are neuroblastoma, lymphoma, leukemia, neurofibroma, hemangioma and fibromatousis [2,15]. The most common sites of metastatic involvement are soft tissues, serosal surfaces, lung, bone marrow, and lymph nodes [19]. In comparison with other soft tissue tumors, lymph node metastasis is frequent in rhabdomyosarcoma and is of a great value in tumor staging [19,20]. The prognosis of embryonal rhabdomyosarcoma has markedly improved following multimodality treatment with excision, radiation therapy, and multidrug chemotherapy [1,4,7,8,20]. This case is worthwhile to report because of extreme rarity of RMS in neonates. Furthermore, its fast growth in less than one month was noteworthy; otherwise, the patient seemed to be normal at birth and the symptoms began one month after. Most of the previously reported congenital embryonal RMS had male dominance, but our patient was female [1,6,16,17]. The radiological examination revealed multiple involvements through the whole body, including subcutaneous lesions and several lymphadenopathies. Although the metastases to bone marrow was established through histologic examination of bone marrow aspiration, it was uncertain whether it has multiple primaries, or from the left extremity and metastasize to other sites. Immunohistochemical staining confirmed the diagnosis, as rhabdomyosarcoma shows positivity for desmin and myoglobin. We examined other markers in this case for ruling out other probable important differential diagnoses. As Synaptophysin, chromogranin, LCA (CD45), CD99, WT1, CD68 and CD1a were negative, besides immunopositivity for muscle markers, Neuroblastoma (Synaptophysin+, chromogranin+, Myogenin-,

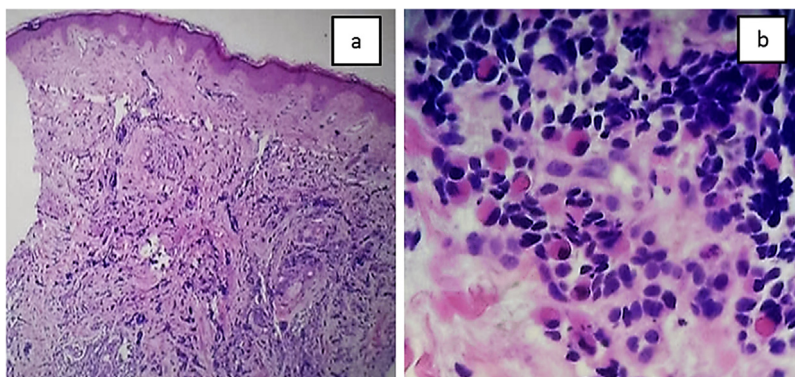


Fig. 3. low power(a) & High power(b) skin lesion microscopy, showing multiple rhabdomyoblasts in this field.

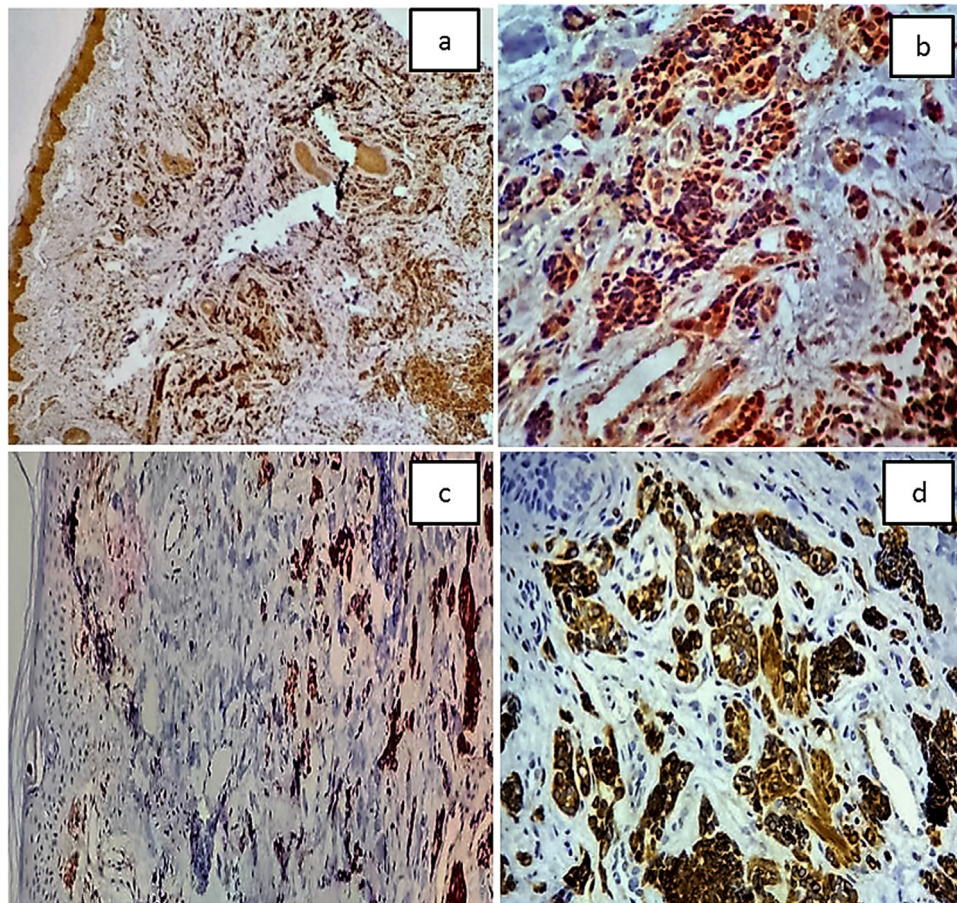


Fig. 4. Immunohistochemical staining of tumoral cells in dermal lesion show strong immunopositivity for Myogenin in low power microscopy(a) and high power (b) and strong positivity for Desmin in low power microscopy(c) and high power (d).

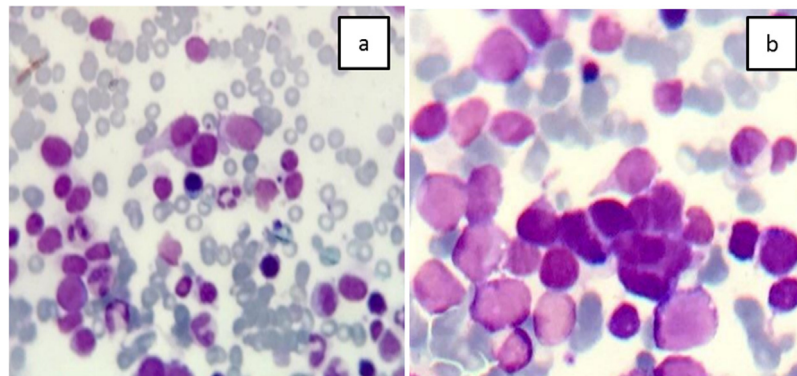


Fig. 5. Bone marrow involvement by rhabdomyosarcoma in high power microscopy (a) One tadpole like tumoral cell is seen in the center of this field in oil power microscopy (b).

Desmin-), Large cell lymphoma(CD45+, B/T cell markers present, desmin-), Acute lymphoblastic leukemia(LCA+, CD99+, Myogenin-, Desmin-), Wilms tumor(WT1+, Myogenin-, Desmin-), Ewing sarcoma (CD99+, Myogenin-, Desmin-) and Langerhans cell histiocytosis(CD68+ and CD1a+) were excluded; hence, our case did not fit any of these diagnoses [18–20].

4. Conclusion

We describe a patient with unusual presentation of multiple rhabdomyosarcoma of a female neonate. it was uncertain whether it has multiple primaries, or from the left extremity and metastasize

to other sites. This rare condition should be considered clinically in the differential diagnosis of neuroblastoma, lymphoma, leukemia, neurofibroma, hemangioma, fibromatousis and mastocytosis. It is important to keep in mind that Rhabdomyosarcoma can be seen as multiple congenital lesions in neonates, therefor necessary diagnostic imunohistochemical staining should be done to the better management of the disease.

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Conflicts of interest

None of the authors have any commercial or financial involvement in connection with this study that represents or appears to represent any conflicts of interest

Consent

Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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References

- [1] G.B. Singh, R. Arora, D. Kumar, M. Jain, V. Puri, Case of Congenital Rhabdomyosarcoma with Review of the Literature, *Case Rep. Otolaryngol.* (2013) 2013, Hindawi Publishing Corporation.
- [2] F.B. Ruymann, W.A. Newton Jr, A.H. Ragab, M.H. Donaldson, M. Foulkes, Bone marrow metastases at diagnosis in children and adolescents with rhabdomyosarcoma. A report from the Intergroup Rhabdomyosarcoma Study, *Cancer* 53 (1984) 368–373.
- [3] C. Rodriguez-Galindo, D.A. Hill, O. Onyekwere, et al., Neonatal alveolar rhabdomyosarcoma with skin and brain metastases, *Cancer* 92 (6) (2001) 1613–1620.
- [4] M. Buyukpamukcu, A. Varan, C. Tanyel, M.E. Senocak, S. Gogus, C. Akyuz, T. Kutluk, N. Buyukpamukcu, Solid tumors in the neonatal period, *Clin. Pediatr.* 42 (2003) 29–34.
- [5] H. Isaacs Jr., Congenital and neonatal malignant tumors: a 28-year experience at Children's Hospital of Los Angeles, *Am. J. Pediatr. Hematol. Oncol.* 9 (1987) 121–129.
- [6] O. Singh, S. Sh. Gupta, V. Upadhyaya, Sh. Sh. Sharma, B.K. Lahoti, K.R. Mathur, Rhabdomyosarcoma of the posterior chest wall in a newborn: a case report, *Cases J.* 2 (2009) 6818.
- [7] G. Moretti, R. Guimarães, K.M. de Oliveira, F. Sanjar, R.L. Voegels, Rhabdomyosarcoma of the head and neck: 24 cases and literature review, *Braz. J. Otorhinolaryngol.* 76 (4) (2010) 533–537.
- [8] A. Damor, B. Patel, Ru. Mapshekar, N. Patel, Congenital rhabdomyosarcoma: a rare axillary presentation, *Int. J. Biomed. Adv. Res.* 3 (2012) 212–214.
- [9] A. Ferrari, M. Casanova, G. Bisogno, I. Zanetti, G. Cecchetto, B. Bernardi, R. Riccardi, P. Tamaro, C. Meazza, R. Alaggio, V. Ninfo, M. Carli, Rhabdomyosarcoma in infants younger than one year old, *Cancer* 97 (10) (2003) 2597–2603.
- [10] C. Kuhnen, P. Herter, I. Leuschner, I. Mentzel, D. Druecke, M. Jaworska, G. Johnen, Sclerosing pseudovascular rhabdomyosarcoma – immunohistochemical, ultrastructural, and genetic findings indicating a distinct subtype of rhabdomyosarcoma, *Virchows Arch.* 449 (2006) 572–578.
- [11] A.O. Cavazzana, O Schmidt, V. Ninfo, O Harms, M. Tollot, M. Carli, L. Treuner, R. Bello, G. Salviati, Spindle cell rhabdomyosarcoma. A prognostically favorable variant of rhabdomyosarcoma, *Am. J. Surg. Pathol.* 16 (1992) 229235.
- [12] R. Kodet, W.A. Newton Jr, A.B. Hamoudi, L. Asmar, D.L. Jacobs, H.M. Maurer, Childhood rhabdomyosarcoma with anaplastic (pleomorphic) features. A report of the Intergroup Rhabdomyosarcoma Study, *Am. J. Surg. Pathol.* 17 (1993) 443–453.
- [13] S.W. Weiss, J.R. Goldblum, Rhabdomyosarcoma, in: S.W. Weiss, J.R. Goldblum (Eds.), *Enzinger and Weiss's Soft Tissue Tumors*, 4th edition, CV Mosby, St Louis, 2001, pp. 785–835, Masson JK, Soule Ell.
- [14] E.H. Soule, M. Geitz, E.D. Henderson, Embryonal rhabdomyosarcoma of the limbs and limb-girdles. A clinicopathologic study of 61 cases, *Cancer* 23 (1969) 1336–1346.
- [15] S.M. Kraft, V. Singh, K.J. Sykes, A. Gamis, M.A. Manalang, J.L. Wei, Differentiating between congenital rhabdomyosarcoma versus fibromatosis of the pediatric tongue, *Int. J. Pediatr. Otorhinolaryngol.* (2010).
- [16] F. Khatami, A. Bazrafshan, M. Monajemzadeh, M. Seyed1, Congenital embryonal rhabdomyosarcoma with prenatal onset, *Iran. J. Pediatr.* 18 (1) (2008) 62–66.
- [17] S.N. Gupta, A.N. Gangopadhyay, S. Khanna, Congenital embryonal rhabdomyosarcoma of the tongue, *Pediatr. Surg. Int.* 5 (4) (1990) 284–286.
- [18] K.R. Cho, J.L. Olson, J.I. Epstein, Primitive rhabdomyosarcoma presenting with diffuse bone marrow involvement. An immunohistochemical and ultrastructural study, *Mod. Pathol.* 1 (1988) 23–28.
- [19] Y. Nishida, S. Tsukushi, H. Urakawa, H. Sugiura, H. Nakashima, Y. Yamada, N. Ishiguro, High incidence of regional and in-transit lymph node metastasis in patients with alveolar rhabdomyosarcoma, *Int. J. Clin. Oncol.* 19 (3) (2014) 536–543.
- [20] C.T. Scheltinga, P. Spronk, J. Rosmalen, M.H. Wijnen, R. Baren, H.K. Merks, Diagnosis and treatment of lymph node metastases in pediatric rhabdomyosarcoma in the Netherlands, *J. Pediatr. Surg.* (2014) 416–419.

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