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Case Report Open Access

Atypical Rhabdoid Teratoid Tumour and Localized Lymphedema in an Infant

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Abstract

Lymphedema is a chronic progressive and multifactorial disorder of lymphatic vessels which is characterised by the regional accumulation of excessive amounts of protein-rich fluid in intersititial space. It may be primary as obstruction of lymphatic drainage due to genetic structural deformity of lymphatic vessels or secondary to infections, surgery, venous insufficiency, trauma and malignancy. Clinical manifestation may vary according to the underlying etiology and duration of pathogenesis.

We want to present a 2 month-old caucasian boy with a pink, slowly growing flaccid, protruding tumoral lesion on his neck that exists since birth. Punch biopsy specimen was consistent with lymphedema. Two weeks after the diagnosis he was taken to emergency service with a sudden onset nausea, vomiting and loss of consciousness, cranial computed tomography performed him and a 5x4 cm in diameter, heterogenous tumor was detected in posterior fossa. Because of accompanying symptoms of hydrocephalus, patient received an urgent operation. Excisional biopsy of the tumour was consistent with stage 4 atypical rhabdoid/teratoid tumor. Unfortunately patient was lost on postoperative day 0.

Atypical rhabdoid/teratoid tumour is a rare and highly aggressive tumour of central nervous system that is usually seen in infancy and early childhood. In our patient neck localized lymphedema was probably associated with cerebellar atypical rhabdoid/teratoid tumor. To our knowledge this is the first case report in the literature.

Keywords: Atypical rhabdoid/ teratoid tumour; Lymphedema

Introduction

Lymphedema is a progressive and multifactorial disease of lymphatic vessels [1]. They may be reduced in number, obliterated or obstructed as a result of primary or secondary diseases [2]. Although infections are reported to be the most common reason of lymphedema all over the world, increasing number of neoplastic diseases and therapy modalities seem to take the first place. Herein we want to present an infant with localized lymphedema which is associated with a very rare central nervous system tumour.

Case Report

A 2 month-old caucasian boy was brought up to our outpatient policlinic, Istanbul Medeniyet University, Department of Dermatovenerology by his family with an asymptomatic, soft, pink, slowly growing tumoral lesion on his neck that exists since birth (Figure 1). He was a term baby and born with spontaneous vaginal delivery with normal birth weight. His growth and developmental parameters were normal according to his age and systemic examination, neurological examinations were also normal. He was first baby of his family. There was no consanguinity between mother and father also both mother and father has no genetic, systemic or dermatological disease.

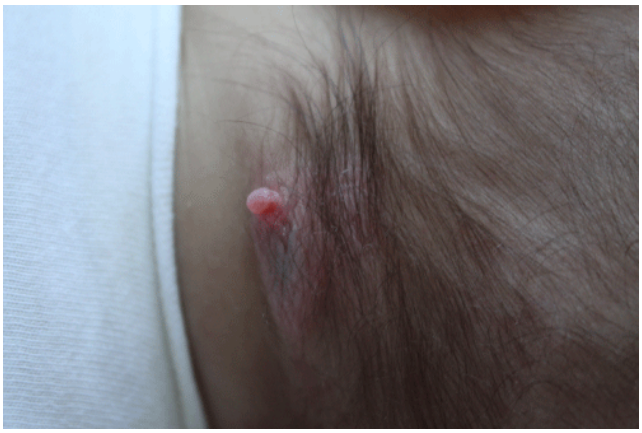

Figure 1: Pink, soft, slowly growing papular lesion on an erythematous verrucous plaque on left occipital region of the baby.

A 3 mm punch biopsy specimen was taken from the pink, fibroma-like papular lesion on his neck. Histopathological examination revealed acanthosis of epidermis and edema associated with increased in fibroblasts, and dilated lymphatic vessels with tickened basal membrane in dermis (Figure 2). Accumulation of mucin was not seen in dermis with PAS-Alcian Blue and Mucin Carmine staining. D2 40 staning was positive in endotelial cells. Two weeks after the diagnosis, he was taken to the emergency service because of a sudden onset

nausea, vomiting and loss of consciousness. A cranial computed tomography performed him and a 5×4 cm in diameter, heterogenous tumour was detected in posterior fossa (Figure 3). Because of accompanying hydrocephalus symptoms he received an urgent cranial operation. Excisional frozen biopsy of the tumour revealed rhabdoid like features in some cells which have eccentric vesicular nuclei and cytoplasmic vacuolization, small blue tumour cells which are infiltrating choroid plexus, PNET -like area of atypical teratoid/ rhabdoid tumour, immunhistochemical staining revealed GFAP expresion of tumour cells which was consistent with stage 4 atypical rhabdoid/teratoid tumor that was lying into bilateral serebellar hemisphere and vermis (Figure 4). Unfortunately he died on postoperative day 0. His parents did not allow performing an autopsy.

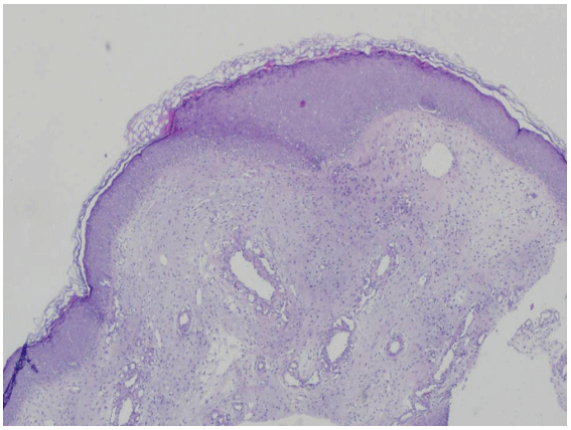

Figure 2: Acanthosis of epidermis and edema associated with increased in fibroblasts and dilated lymphatic vessels with tickened basal membrane in dermis.

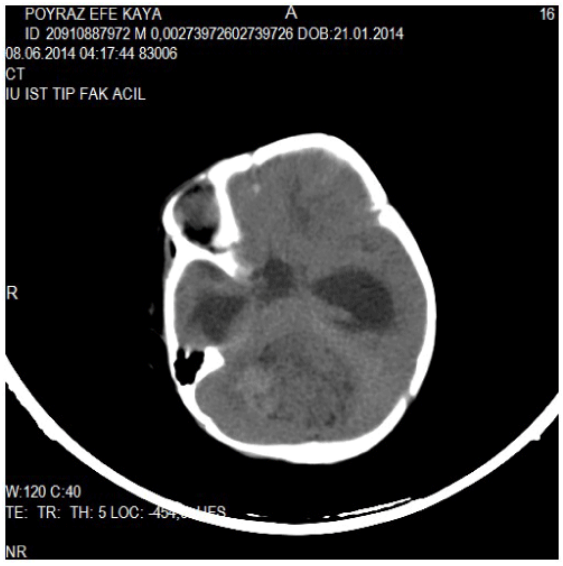

Figure 3: A 5×4 cm in diameter, heterogenous tumour in posterior fossa.

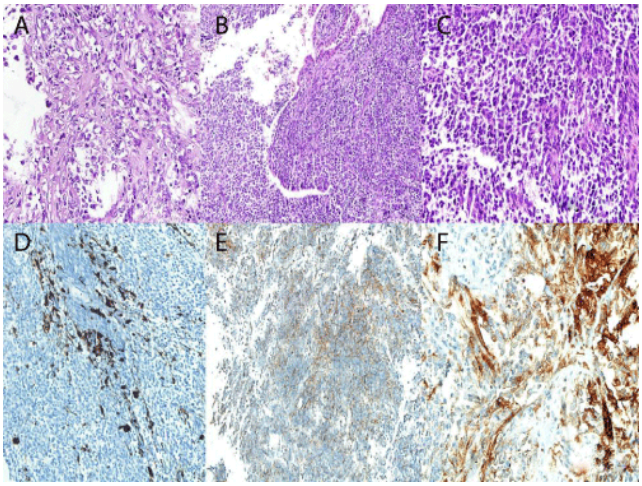

Figure 4A: Rhabdoid like features in some cells which have eccentric vesicular nuclei and cytoplasmic vacuolization (H/ E X 400). **Figure 4B:** The "small blue" tumour cells which are infiltrating choroid plexus (H/E X 200). **Figure 4C:** PNET -like area of atypical teratoid /rhabdoid tumour (H/ E X 400). **Figure 4D:** Immunhistochemical GFAP expresion of tumour cells (GFAPx200). **Figure 4E:** Immunhistochemical synaptophysin expresion of tumour cells (Synaptophysin x 200). **Figure 4F:** Membranous and cytoplasmic EMA expression of tumour cells (EMA x 400).

Discussion

Chronic lymphedema is a difficult disorder to treat which could be primary or secondary to many localized or systemic disorders [2]. Primary form is usually seen with intrinsic abnormalities of lymphatic vessels and may be classified in 3 groups according to the age that lesions first appeared or in 2 groups according to the etiology [3]. Congenital primary lymphedema presents at birth or before age 2 and is usually associated with hereditary disorders such as Milroy's disease, Lymphoedema- distichiasis syndrome, Noonan Syndrome, Turner Syndrome, yellow nail syndrome, Hennekam Syndrome [2-4]. The other form of primary lyphedema is acquired primary lymphedema and this form is usually associated with intraluminal or intramural lymphangio-obstructive edema of proximal or distal lymphatics and obstruction of the lymph nodes [3]. Secondary lymphedema may be associated with several pathological process which lead to acquired obstruction or obliteration of lymphatics such as infections, malignancies, inflammation, obesity, granulomatous diseases, vascular diseases and trauma [2]. Malignancy related secondary lymphedema is usually seen in adults and occurs therapy associated either as a result of surgery, radiotherapy or both of them. Also neoplastic infiltration of the lymphatic vessels, lymph nodes (metastases) and lymphatic ducts (external compression or carcinomatous lymphangitis) are the most common causes of malignancy related lymphedema [2,3,5]. Best known samples are breast cancer associated lymphedema of the upper extremity and prostate cancer associated lymphedema of lower extremity and pubic region [3,6,7]. In our case neck localized fibroma like lesion was consistent with lymphedema histopathologically and presented on a very unusual localisation. We think in our case lymphedema was associated with intracranial neoplasm due to the

lesion's projection. Intracranial lesion was detected in left serebellar region in posterior fossa and cutaneous lesion of lymphedema was also localized on left occipital region. This presentation may be associated with chronic pressure of this large tumour to logistic lymphatics and sceletal structures. Histopathologically lymphedema is characterised by swollen and seperated collagen fibres, perivascular infiltartion of mononuclear cells, thickened and fibrosen lymphatic vessels and fibrosis. The number of blood vessels greatly increases [2]. In our case histopahological examination of skin lesion located on neck revealed increased fibroblasts and dilated lymphatic vessels with tickened basal membrane in dermis.

Atypical Teratoid/Rhabdoid Tumors (AT/RT) is a very rare and highly malignant embryonal tumor of central nervous system (CNS) and predominantly occurs in young children under 3 years old [8-11]. Characteristic feature of AT/RT is aberrations of the SMARCB1 (hSNF5/INI1) gene [8]. It was first decribed by Roker in 1996 and was introduced to the WHO brain tumor classification in 2000 [8]. SMARCB1 gene mutation could not be performed to our patient because of technical disability. Histopathologically, AT/RT is characterized by rhabdoid tumour cells, which have vesicular nuclei, large nucleoli, and cytoplasmic filamentous inclusions. In our case myxoid degeneration, increased fibroblastic activity and dilatation in vascular structure in dermis were seen histologically. Systemic or intratechal chemotherapy, radiotherapy and surgery are most common treatment choices of AT/RT [11]. Our patinet received an urgent surgery because of accompanying symptoms of hidrocephalus and he was diagnosed as AT/RT after the operation but he was lost in intensive care unit on postoperative day 0. Cutaneous manifestation of AT/RT has been reported as an axillary mass before but to our knowledge AT/RT associated lymphedema has not been reported in the literature before [12]. We want to remind probable malignancy in isolated cases of localized lymphedema with this case report.

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