Sclerosing Rhabdomyosarcoma of the Lip in a Child; A Case Report

Çocuk Hastada Dudak Yerleşimli Sklerozan Rabdomyosarkom; Olgu Sunumu

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Abstract
Rhabdomyosarcoma is the most common malignancy of soft tissue in childhood and adolescence. It has been traditionally classified into 3 main subtypes: embryonal, alveolar and pleomorphic. However a new, rare subtype called Spindle cell/sclerosing Rhabdomyosarcoma has been defined recently and classified as the fourth variant by the World Health Organization in 2013. We report a case of a 4 year-old girl who had a solid mass in her lower lip with a size of 1cm. In the histopathologic examination the lesion was predominantly composed of mitotic active spindle-shaped cells with hyperchromatic nuclei in a sclerotic stroma and some rhabdomyoblastic cells with eosinophilic cross-striated cytoplasm. All of these cells were stained positive with Vimentin, Desmin, Myogenin, CD 99 and CD 56. Therefore the lesion was diagnosed as Spindle cell/sclerosing Rhabdomyosarcoma. This case reminds pathologists and the clinicians that this rare entity should be remembered in the differential diagnosis of spindle-cell tumors in childhood.
Keywords: Childhood, Lip, Sclerosing rhabdomyosarcoma

Özet
Rabdomyosarkom; çocukluk ve adolesan çağda yumuşak dokunun en sık görülen malignitesidir. Klasik olarak embriyonel, alveoler ve pleomorfik olarak 3 ana subtipe ayrılmaktadır. Ancak son olarak, yeni ve nadir görülen, İğsi hücreli/Skrerozan Rabdomyosarkom olarak tanımlanan bir subtipi, 2013 yılında Dünya Sağlık Örgütü 4. varyant olarak sınıflamaya dahil etmiştir. Yazımızda alt dudağında 1cm çaplı solid bir kitlesi olan 4 yaşında bir kız çocuğunu tartışmaktayız. Bu kitle histopatolojik olarak sklerotik stromada çoğunlukla mitotik olarak aktif, hiperkromatik nükleuslu, iğsi- hücreli ve bir kısmı da eozinofilik, çizgili sitoplazmalı rabdomyoblastik görünümlü, Vimentin, Desmin, Myogenin, CD 99 ve CD 56 ile pozitif boyanan hücrelerden oluşmaktaydı. Bu nedenle olgu İğsi hücreli/Sklerozan Rabdomyosarkom olarak tanı aldı. Bu olgu patolog ve klinisyenlere çocukluk çağının İğsi hücreli tümörlerinin ayırıcı tanısında nadir bir varyant olarak akılda bulundurulması gerektiğini vurgulamak amacıyla sunulmuştur.

Anahtar Kelimeler: Çocukluk çağı, Dudak, Sklerozan rabdomyosarkom

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Introduction

Rhabdomyosarcoma (RMS) is the most common malignancy of soft tissue in childhood and adolescence. RMS has been traditionally classified into 3 main subtypes: embryonal, alveolar and pleomorphic (1). However, Mentzel and Katenkamp first reported a distinctive variant of RMS called Sclerosing, Pseudovascular Rhabdomyosarcoma; characterized by prominent hyaline sclerosis and a pseudovascular growth pattern in 2000 (2). In 2002, Folpe et al. presented 4 new cases in adults with similar features and they entitled this variant as Sclerosing rhabdomyosarcoma (3). This new variant was included in the World Health Organization (WHO)-2013 classification as Spindle cell/sclerosing Rhabdomyosarcoma (SSRMS) (4). This is an uncommon variant of RMS accounting for 5-10% of all cases of RMS. It can be seen in both children and adults. In children the most common site of involvement is the paratesticular region, however deep soft tissues in head and neck are the foremost localizations in adults (4). Here we report a case of SSRMS of a child, that was originated from an unusual region; the lower lip of her mouth.

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Case

A 4-year-old girl was admitted to our hospital with a painless mass on the lower lip of her mouth. This mass had a rapid growth during 2 months and had a diameter of 1cm. Her medical history was unremarkable; however her father died of Non-Hodgkin Lymphoma 3 years ago. Radiologically her mass was round, solid but fixed on the lower lip as if it was a lesion of the minor salivary glands. No other masses or lymph nodes were declared. The material surgically resected had a grey-white, smooth surface and solid, whorled cut surface. All of the material were processed and examined histologically. In the hematoxylin-eosin (HE) slides (Figure 1 and Figure 2), there were many spindle-shaped atypical tumor cells admixed with oval, polygonal shaped cells with eosinophilic cytoplasm infiltrating the striated muscles, connective and adipose tissues of the lip. These cells trend to form fascicles, bands, small nests in an abundant collagenous stroma showing a prominent hyaline sclerosis and a focally chondroid-like appearance. Mitotic activity was high up to 10 mitotic figures per 10 high power fields and tumor necrosis was also present focally. Immunhistochemically, tumor

cells stained positive for Vimentin, Desmin (Figure 3), Myogenin (Figure 4), CD 99 and CD 56. With all of these findings the tumor was diagnosed as SSRMS. However an additonal re-excision had to be planned as the material had infiltrative, tumor positive surgical margins.

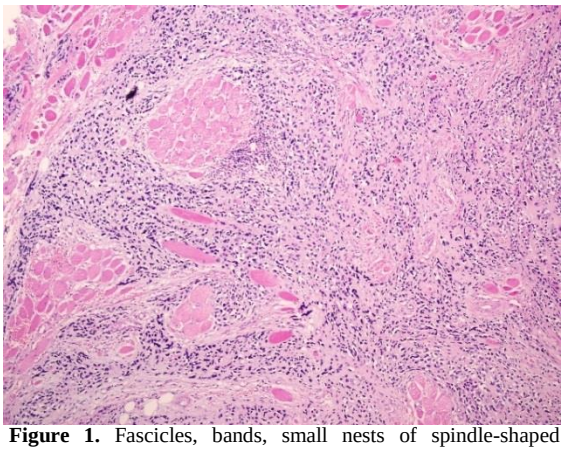

Figure 1. Fascicles, bands, small nests of spindle-shaped atypical tumor cells infiltrating the striated muscles and connective tissues in a sclerotic collagenous stroma. (HE, x100)

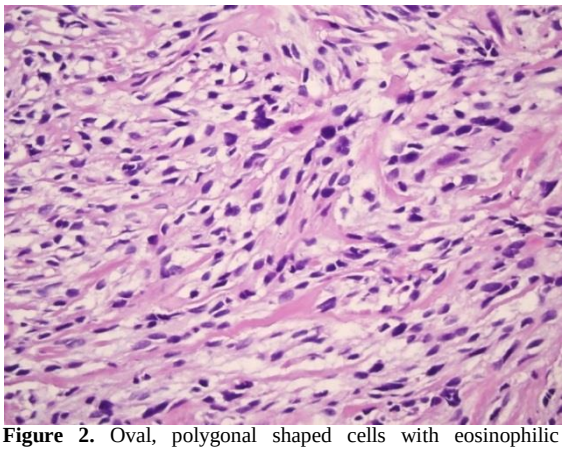

Figure 2. Oval, polygonal shaped cells with eosinophilic cytoplasm scattered throughout the tumor (HE, x400)

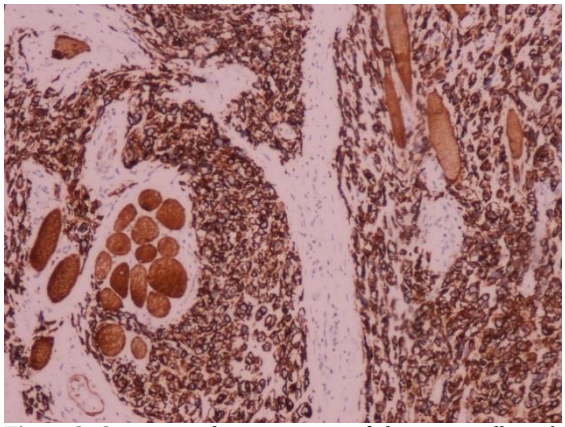

Figure 3. Strong cytoplasmic staining of the tumor cells with Desmin (Desmin, x200)

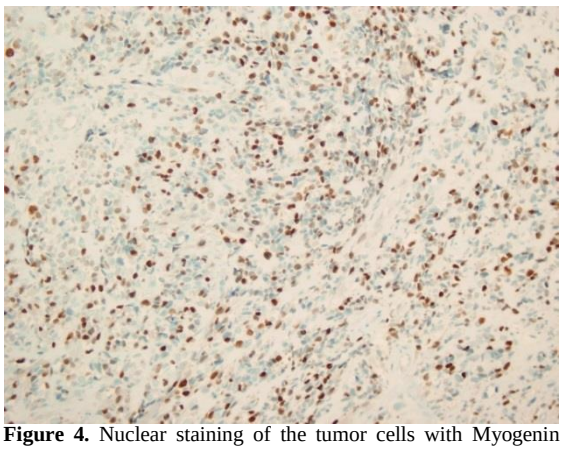

Figure 4. Nuclear staining of the tumor cells with Myogenin (Myogenin, x200)

Discussion

In 1992, spindle cell RMS was first defined as a variant of embryonal RMS in the pediatric population by the German-Italian Cooperative Sarcoma Study. They presented 21 cases, which were mostly involved in the paratesticular region, i.e., the head and neck. They stated that this subtype had a low malignant potential, therefore it should be distinguished from the classical forms of embryonal RMS (5). In 1998, Rubin et al. presented some cases of spindle cell RMS in adults having a more aggressive clinical course (6). Later Mentzel (2) and Folpe (3) defined a variant of RMS, with a sclerosing stroma and spindle cells named as Sclerosing RMS.

So RMS comprises a heterogeneous group of lesions that contain skeletal muscle differentiation. Subtyping of RMS is important as they differ in clinical, histological, molecular and prognostic features from the other subtypes. This is important especially in children and adolescents, where prognostically relevant subgroups are defined and treated by different protocols (7). WHO recently classified RMS into 4 groups including embryonal, alveolar, pleomorphic and Spindle cell/sclerosing subtypes. Embryonal and Spindle cell/sclerosing subtypes arising especially in children and adolescents especially have excellent prognosis. In contrast, alveolar and pleomorphic subtypes have a worse clinical outcome (4,7).

SSRMS is the least common variant, accounting for 5-10% of all cases of RMS. It affects both children and adults, with a male to female ratio of 6:1. In pediatric population, this variant arises predominantly in the paratesticular area, while in adults >50% of cases affect the deep soft tissues in the head and neck. In both age groups the limbs are the most affected sites (4). Lips (like in our case) are rare regions for SSRMS to originate from in childhood. Microscopically, SSRMS is composed of predominant population of spindle shaped tumor cells with ovoid or elongated nuclei, vesicular chromatin, inconspicuous nucleoli and scant

eosinophilic cytoplasm. Rare to scattered rhabdomyoblasts can be seen usually throughout the tumor. Nuclear atypia, hyperchromasia and mitotic figures are common. The tumor cells form nests, fascicles, and trabeculae in a very sclerotic stroma. And they are expected to be stained positive with Desmin, Myogenin, Vimentin, CD 99 and WT-1 (4,7). The treatment recommended is complete tumor resection combined with systemic chemotherapy. The VAC (Vincristine, Actinomycin D and Cyclophosphamide) regimen is one of the recommended therapeutic options. More than half of these tumors recur or progress locally or distantly without bone marrow invasion (8).

Our patient was a child with a mass located in an unusual area; the lower lip of her mouth. Predominant spindle cells some of which had hyperchromatic nuclei with eosinophilic cytoplasm formed the tumor fascicles and trabeculae. Tumor cells were positive stained with Desmin, Myogenin, Vimentin and CD 99 as expected.

In the literature, SSRMS in adults have been reported in many articles with a worse prognosis than the pediatric group so far; however SSRMS in the mouth region in children is rather rare and the prognosis of it is still unclear. As more cases are recognized and reported, it will be possible to define the clinicopathologic and prognostic features of this rare entity accurately and it will be possible to find more optimal treatment strategy to improve the prognosis.

Informed Consent: Written informed consent was obtained from patient who participated in this case (14.07.2016).

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