Journal of Pediatric Surgery (2009) **44**, E5–E8

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Journal of Pediatric Surgery

www.elsevier.com/locate/jpedsurg

Multifocal leiomyosarcomatosis in a 6-year-old child with epidermodysplasia verruciformis and immune defect

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Received 9 November 2008; revised 2 April 2009; accepted 4 April 2009

Key words:
Multifocal leiomyosarcomatosis;
Epidermodysplasia verruciformis;
Immune defect

Abstract Leiomyosarcoma and epidermodysplasia verruciformis are rarely encountered in children. The association of either leiomyosarcoma or epidermodysplasia verruciformis with immune deficiency has previously been documented. A 6-year-old girl, who has had multifocal leiomyosarcomatosis after the previous diagnoses of epidermodysplasia verruciformis and immune defect, represents an interesting association of these features.

Leiomyosarcoma is a malignant smooth muscle tumor that accounts for 2% to 4% of soft tissue tumors in children [1,2]. Unlike the adult form, pediatric leiomyosarcoma arises less commonly in the retroperitoneum and genitourinary tract. The most frequent primary sites are the gastrointestinal tract (especially stomach), head, and neck [1]. Multifocal leiomyosarcomatosis may be associated with congenital and acquired immune deficiencies [3].

Epidermodysplasia verruciformis (EV) is a rare disorder, which is characterized by persistent human papillomavirus (HPV) infection with an autosomal recessive inheritance. The clinical diagnostic features are lifelong eruption of pityriasis versicolor-like macules; flat, wartlike papules; and the development of cutaneous carcinomas [4]. The development of EV has also been defined in immunodeficiency states [4,5].

Herein, we report a 6-year-old girl with multifocal leiomyosarcomatosis associated with EV and an ill-defined immune defect.

1. Case report

A 6-year-old girl was admitted for abdominal pain, bloody stool, and weight loss of 6-month duration. She experienced the occurrence of pruritic skin lesions since 6 months of age. She also had recurrent respiratory tract infections and episodes of diarrhea since 3 years of age. She was previously hospitalized for several times for these

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doi:10.1016/j.jpedsurg.2009.04.011

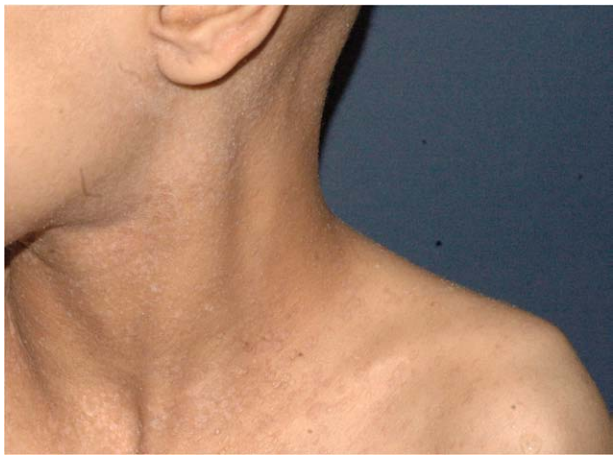

Fig. 1 Multiple discrete verruciform papules and macules on the V of the neck and shoulders.

complaints. She had multiple sites of pulmonary consolidation and pretracheal lymphadenopathy on chest computed tomography when she was 4 years of age.

One month before her current admission, she was admitted to another hospital for abdominal pain and bloody stool. Abdominal ultrasound and abdominal magnetic resonance imaging showed multiple contrast enhancing nodules in both lobes of the liver, a thickened gallbladder wall, and a tumorlike lesion 2 cm in diameter.

On admission to our institution, she appeared chronically ill. Her body weight was between the 3rd and 10th percentiles. There was no family history of consanguinity or cancer.

Physical examination revealed hepatomegaly. Dermatologic examination showed widespread light brown to skin-colored, slightly hyperkeratotic macules and flat verruciform papules on the neck, trunk, and the forearms that were clinically consistent with EV (Fig. 1). The papules coalesced to form plaques on the antecubital areas (Fig. 2). Her hair and nails were normal. The skin biopsy showed hyperkeratosis in the epidermis and minimal acantosis. There was swelling and basophilic cytoplasm of keratinocytes containing basophilic

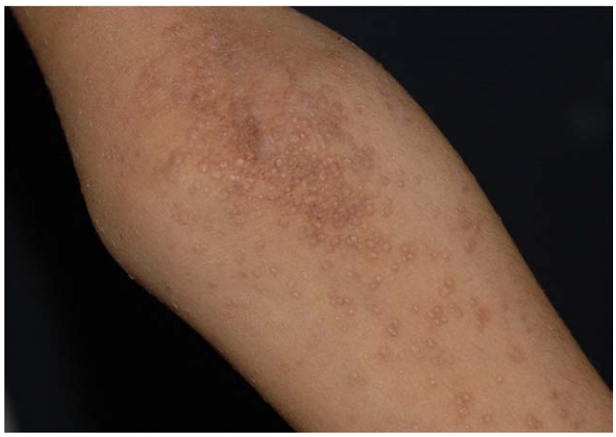

Fig. 2 Papules forming plaques on flexor extremities.

hyaline granules within it. The immunofluorescence examination of IgA, IgG, IgE, IgM, C1q, C3, C4, and fibrinogen was negative. These finding were reported as EV pathologically. However, the PCR examination for HPV serotype from the superficial scaly specimen of lesions was negative.

Laboratory findings including total blood count, urinalysis, and blood biochemistries were within normal limits. The erythrocyte sedimentation rate was 100 mm/h (reference range, 0-20). Serologic testing for HIV, cytomegalovirus, and Epstein-Barr virus (EBV) in blood was performed and revealed positive anti-cytomegalovirus IgG and anti-EBV virus-capsid antigen (VCA) IgG antibodies. Abdominal ultrasonography and computed tomography showed multiple nodules in both lobes of the liver. The wall of the gallbladder was thickened, and multiple heterogeneous solid lesions were seen within it. There were 2 solid tumorlike lesions in the left retroperitoneal region just superior to the left kidney, as well as a solid lesion with a diameter of 2 cm within the loops of intestines in the right lower quadrant (Fig. 3). Chest computed tomography revealed multiple bilateral metastatic nodules within the lungs.

Immunologic investigations including immunoglobulin levels, lymphocyte subsets, and blastic transformation were performed. The positive findings were elevated levels of IgE and IgG and reduced percentage of CD4-positive cells (14%) with a CD4/CD8 ratio of 0.4. However, the total number of CD4-positive cells was within normal limits. Lymphocyte proliferation by various mitogens was reported as mildly to moderately low.

A needle biopsy from one of the liver lesions was performed. Histopathologic examination of the specimen revealed a smooth muscle tumor, but it was not possible to decide whether it was malignant. An explorative laparotomy was performed that showed 2 nodules with the largest diameters of 3 and 3 cm, respectively, located just superior to the left kidney. They were totally excised. The gallbladder was enlarged; its wall was thickened and firm to palpation. A

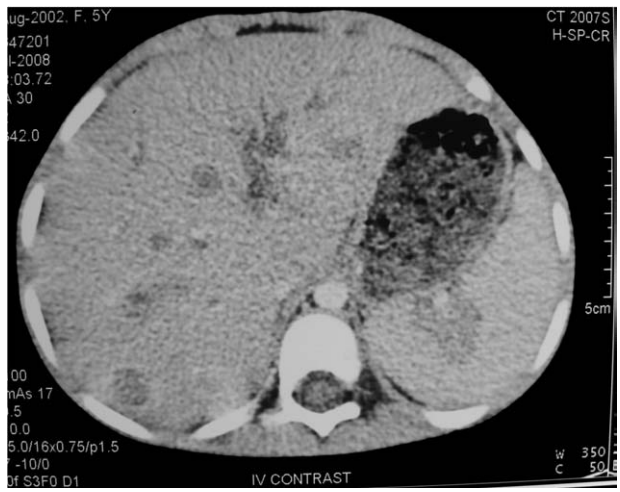

Fig. 3 Abdominal computed tomography revealing multiple nodules in the liver.