CASE REPORT

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A Rare Coexistence of Turner Syndrome and Mycosis Fungoides: A Case Report

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What is already known on this topic?

Turner syndrome is linked to an increased prevalence of several autoimmune diseases and certain cancers, especially malignant melanoma, and nervous system and gastrointestinal malignancies. Mycosis fungoides, the most common primary cutaneous T-cell lymphoma, affects adults and children with slow progression that requires careful monitoring.

What this study adds?

This is the first published case of an 11-year-old girl with both Turner syndrome and mycosis fungoides. It highlights the importance of thorough dermatologic evaluation in Turner syndrome patients, especially for atypical skin lesions, suggesting mycosis fungoides as a potential differential diagnosis.

ABSTRACT

Turner syndrome (TS) is the most common sex chromosome abnormality among females, characterized by short stature, hypergonadotropic hypogonadism, congenital heart anomalies, and an increased risk of autoimmune diseases. Although TS does not typically increase the absolute risk of malignancy, specific cancers, such as those affecting the nervous system and gastrointestinal tract and malignant melanoma, may occur more frequently. Mycosis fungoides (MF) is the most common type of primary cutaneous T-cell lymphoma, generally affecting otherwise healthy adults but also seen in children and adolescents. We report an 11.2-year-old girl with TS presenting with substantial weight gain and short stature. Clinical examination revealed characteristic TS features and karyotype analysis confirmed mosaic TS. Following growth hormone (GH)

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therapy, the patient developed persistent, erythematous, itchy skin lesions diagnosed as folliculotropic MF. GH therapy was discontinued, and topical steroids controlled the skin lesions effectively. MF in TS is very rare and unexpected, especially in a child. This is the first reported case of MF in a child with TS. This case highlights the importance of carefully evaluating skin lesions in patients with TS and suggests considering MF as a differential diagnosis.

Keywords: Turner syndrome, mycosis fungoides, malignancy, primary cutaneous T-cell lymphoma

Introduction

Turner syndrome (TS) is the most common sex chromosome abnormality among females, caused by the complete or partial absence of one of the X chromosomes**,** which is characterised by short stature, hypergonadotropic hypogonadism, congenital heart anomalies, and an increased risk of autoimmune disease (1). Although the absolute risk of malignancy has been reported not to increase in TS, some specific types of cancer, such as nervous system malignancies, gastrointestinal tract malignancies, or malignant melanoma, have been suggested to occur more frequently in patients with TS (2,3).

Mycosis fungoides (MF) is the most common type of primary cutaneous T-cell lymphoma. Adults in their fifth decade are predominantly affected by the condition, but it should be remembered that MF is also the most common cutaneous lymphoma in children and adolescents, albeit very rarely (4). MF typically exhibits an indolent course limited to the skin in the early stages when MF presents with skin lesions as patches or plaques. However, it may involve visceral organs in a small number of patients in advanced stages, usually associated with tumoral skin lesions. Epidermotropic tumor infiltration, atypical T-cell proliferation, and a cerebriform appearance are typical histopathologic manifestations of MF (5,6).

To the best of our knowledge, the coexistence of TS and MF has yet to be reported. Therefore, we report the clinical findings and follow-up of an 11-year-old girl with TS who developed MF.

Case Report

An 11.2-year-old girl was admitted to the outpatient clinic due to her parents' concerns about substantial weight gain and short stature. Her family noticed she was shorter than her peers and gained about 10 kg in the last two years. She was born at the 32nd week of gestation, from the first pregnancy of a 31-year-old mother. Her birth measurements were within normal range. The parents were nonconsanguineous, and the family history was uneventful. Her postnatal development had been entirely normal.

Her anthropometric measurements and pubertal status at the time of referral are shown in Table 1. A plethoric face and low posterior hairline were observed during the physical examination. Notably, she had acanthosis nigricans on the nape and purple striae on her thighs. Moreover, the shortening of both 5th metacarpals was remarkable. Apart from these findings, systemic examination was otherwise normal, and blood pressure was within the appropriate range for the patient's gender and height.

Given the suspicion of Cushing's syndrome, investigations were performed and the results of the dexamethasone suppression test, 24-hour urinary cortisol, and midnight salivary cortisol levels were all found to be normal. However, impaired glucose tolerance was detected during the oral glucose tolerance test, leading to the administration of metformin treatment. Furthermore, other laboratory examinations, including luteinizing hormone, follicle-stimulating hormone, and estradiol, were 2.21 mIU/mL, 12.18 mIU/mL, and 12.43 pg/mL, respectively (Table 1). These results suggested a diagnosis of TS. The karyotype analysis supported our suspicion, revealing compatibility with mosaic TS [45, X/46, X,i(Xq)/46, XX(8/3/49)]. Following the diagnosis of TS, growth hormone (GH) therapy was initiated. However, in the third month of GH treatment, the patient developed persistent, itchy, erythematous, follicular papules and plaques on her back, chest, axilla, and nape of the neck (Figure 1). It was learned that these lesions had recurred irregularly over the last three years and had improved with the use of short-term topical corticosteroid creams, as suggested by a dermatologist. There was no aggravation of the lesions following GH therapy. Aside from these skin lesions, the patient had no personal history of atopy, and familial atopy history was also unremarkable. A biopsy and histopathological examination was performed from a plaque lesion and revealed CD4+ MF with epidermotropic and adnexotropic characteristics (Figure 2). GH therapy was discontinued and topical steroid therapy was initiated. No additional treatment was required during the follow-up period as the initial and newly developed skin lesions remained well-controlled with topical steroids.

Discussion

MF usually affects adults around 50 years old, with a slight male predominance (7). However more than 5% of patients are diagnosed in childhood (8). We report a rare case of MF, a cutaneous malignancy uncommon in childhood, developing in an 11-year-old girl with TS. A wide variety of clinical presentations in patients with MF have been reported, including follicular papules, patches, indurated plaques, hypopigmented,

Table 1. Clinical and laboratory findings of the patient

	At time of referral	Last examination
Anthropometric measurements		
Age (years)	11.2	12.2
Weight kg/(SDS)	40.5/(0.2)	53.6/(0.9)
Height cm/(SDS)	132.8/(-2.1)	138.1/(-2.5)
BMI kg/m^2/(SDS)	22.8/(1.4)	28.1/(2.3)
Puberty stage (Tanner)	B2P1	B3P2
Bone age (year)	12	12.5
PAH cm (SDS)	144	146.6
Target height cm (SDS)	159 cm (-0.69)	
Hormonal profile		
LH (mIU/mL)	2.2	5.9
FSH (mIU/mL)	12.2	12.2
E2 (pg/mL)	12.4	14
AMH (ng/mL) (NR 0.62-11)	0.36	
HbA1C (%)	5.1	5.2
	OGTT- glucose (mg/dL) 0.' : 116 30.' : 131 60.' : 125 90.' : 121 120.': 144	**OGTT- insulin (µU/mL)** 0.' : 29 30.' : 168 60.' : 6 90.' : 9 120.': 36
Imaging		
Echocardiogram	Bicuspid aorta	
Renal US	Normal	
Pelvic US	R ovary 2.1 mL, L ovary 1.2 mL, uterus 3.8 mL	

SDS: standard deviation score, BMI: body mass index, PAH: pulmonary arterial hypertension, LH: luteinizing hormone, FSH: follicle-stimulating hormone, E2: estradiol, AMH: anti-Müllerian hormone, OGTT: oral glucose tolerance test, US: ultrasound

Figure 1. Erythematous plaque on (a) the nape and (**b**) the axilla

hyperpigmented, acneiform (comedones, cysts), or keratosis pilaris-like lesions (9). However, it has been reported that skin changes, including hypopigmentation, hyperpigmentation, and alopecia may accompany TS with a frequency of 5% (1). These skin changes in TS may be challenging to diagnose in the early stages, and may mimic various benign skin lesions like eczema, psoriasis, or other non-neoplastic skin disorders. Therefore, the diagnosis of MF, in this girl with late diagnosed TS, was surprising and unexpected. The clinical course of MF is usually chronic but indolent, characterized by a slow and gradual progression over years or even decades, from patches to more infiltrated plaques and then rarely to tumors (7).

MF may simulate a diverse range of benign inflammatory skin disorders both clinically and histopathologically (10). The differential diagnosis of the presented patient's skin lesions initially included psoriasis and pityriasis rosea; in lesions with follicular accentuation, keratosis pilaris, pityriasis rubra pilaris, and adnexotropic MF were considered. Histopathological examination of biopsied skin lesions revealed a thick band-like lymphocytic infiltrate in the superficial dermis and the presence of nonspongiotic epidermotropism, which is typically described in MF, helping to exclude other dermatoses (Figure 2a). The predominance of CD4 positive lymphocytes in both intraepidermal and dermal infiltration further supported the

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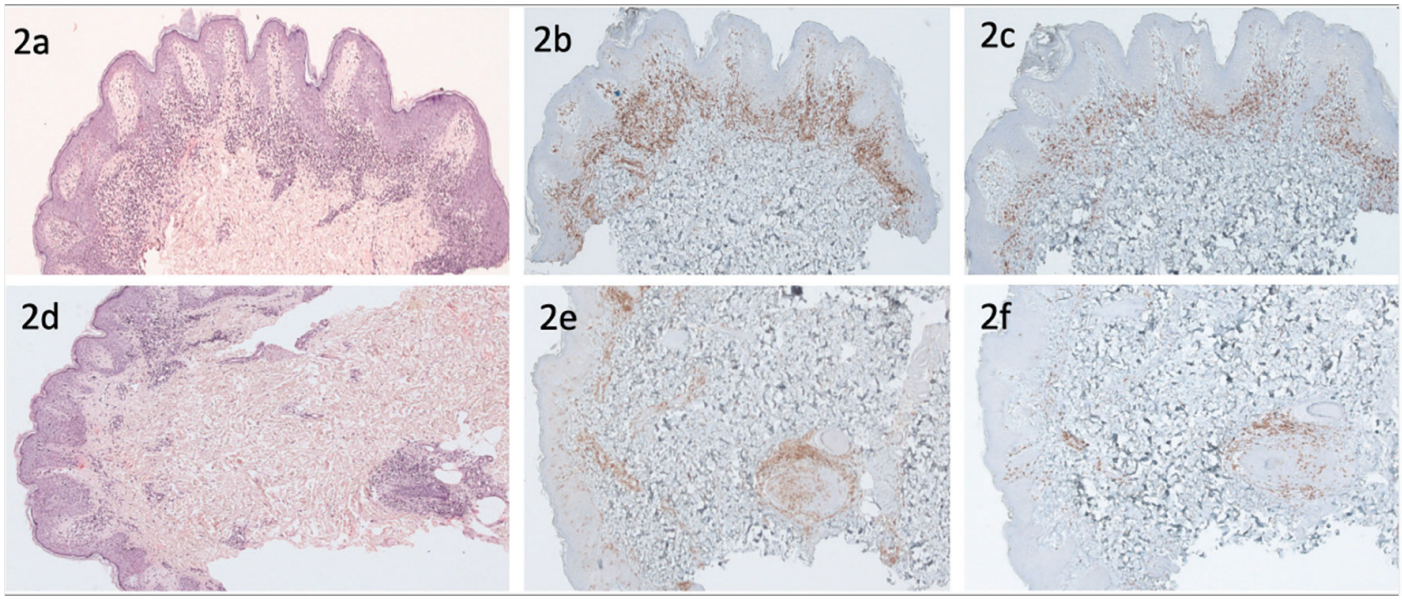

Figure 2. **(a)** Lymphocytic infiltration in the superficial dermis, with a band-like pattern, intermittently contacting the epidermis, in the biopsy taken from the left axilla (H&E, x50), **(b)** Predominance of CD4 staining within the infiltration (CD4, x50), **(c)** CD8 staining of the same specimen (CD8, x50), **(d)** In the biopsy taken from the left side of the trunk, a weaker infiltration is observed in the superficial dermis, while a lymphocytic infiltration around follicles and skin appendages is noticeable in the deeper layers. (H&E, x50), **(e)** Predominant CD4 positivity within the infiltration (CD4, x50), **(f)** Sparse reaction of CD8 staining in dermal lymphocytes (CD8, x50)

diagnosis of MF (Figure 2) (10). Furthermore, biopsies taken from the patient's trunk showed not only epidermotropism but also folliculotropism, supporting the diagnosis of folliculotropic MF.

Due to the presence of recurrent lesions over the last three years, we did not consider that the 3-month GH treatment might be related to MF in our patient. However, given the possibility of increased insulin-like growth factor-1 receptor expression with GH treatment (10), we decided to discontinue the hormonal therapy.

It is known that TS patients have a higher incidence of autoimmune diseases than the general population. Most MF patients are otherwise healthy but other cutaneous or systemic lymphomas have been reported to occur in nearly 7% of patients (11). Furthermore, recent research has indicated an increased prevalence of autoimmune disorders, including inflammatory bowel disease, systemic lupus erythematosus, and type 1 diabetes mellitus, among individuals with MF. This association is believed to stem from T-cell dysregulation (12). The etiology of MF in TS may also be associated with autoimmune dysregulation.

The hormonal abnormalities and treatments in TS may influence the risk of hormone-related malignancies, and the underlying chromosomal abnormality itself may also affect cancer risk (13). Ji et al. (14) reported that patients with TS have an increased risk of solid tumors, particularly malignant melanoma and central nervous system tumors. Another study by Viuff et al. (3) reported that patients with TS with the 45, X karyotype had a two to fivefold increased risk of benign CNS tumors, colorectal malignancies, and malignant melanoma, while TS women with the 45,X/46,XX karyotype had an increased risk of tongue cancer. In the latest guideline, the International Turner Syndrome Consensus Group recommended an annual skin assessment to identify dangerous lymphoedema, dermatitis, infections, autoimmune skin conditions and skin neoplasms, and appropriate evaluation and treatment by a dermatologist, if indicated (15). However, the subtype of neoplastic skin condition was not specified in this guideline. This case also supports an annual assessment by a dermatology specialist which may be beneficial, especially for the presence of skin lesions in cases with TS.

The present report, which describes the development of MF in a girl with TS, contributes to the existing literature concerning TS because, to the best of our knowledge, this is the first published report of the coexistence of MF and TS.

Conclusion

In conclusion, an 11-year-old girl with MF and mosaic TS is described. We believe this to be the first reported instance of this combination. Considering the possible increased risk for malignancies in patients with TS, a thorough evaluation of skin lesions is crucial. We suggest that MF should also be considered as a possible differential diagnosis. A dermatologic assessment may be necessary to confirm the diagnosis and guide appropriate treatment.

Ethics

Informed Consent: Informed consent was granted by the parents of the patient for publication.

Footnotes

Authorship Contributions: Surgical and Medical Practices: Tugba Atci, Sule Oztürk Sari, Can Baykal, Asli Derya Kardelen, Firdevs Bas, Concept: Esin Karakilic-Ozturan, Melek Yildiz, Design: Ozge Bayrak Demirel, Data Collection and Processing: Ozge Bayrak Demirel, Analysis or Interpretation: Sule Ozturk Sari, Can Baykal, Sukran Poyrazoglu, Feyza Darendeliler, Literature Search: Ozge Bayrak Demirel, Esin Karakilic-Ozturan, Melek Yildiz, Writing: Ozge Bayrak Demirel, Tugba Atci, Firdevs Bas.

Conflict of Interest: One author of this article, Feyza Darendeliler, is a member of the Editorial Board of the Journal of Clinical Research in Pediatric Endocrinology. However, the reviewers evaluating this manuscript were blinded and were from different institutions. She was not involved in the editorial review of this manuscript to avoid prejudice that may disrupt impartiality. The other author declares no conflict of interest.
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