Systemic lupus erythematosus in a patient with Turner syndrome*

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Dear Sir,

Turner syndrome (TS), one of the most common sex chromosome disorders, is caused by numeric or structural abnormalities of the X chromosome. It occurs in approximately 1 in every 2,500 newborns with female phenotype. The karyotypes are nonmosaic or mosaic, including 45,X, 46,X,del(Xp), 46,X,i(Xq), 45,X/46,XX, 45,X/46,XrX, 45,X/46,XY, 45X/47XXX. 45,X is a classical type. TS is associated with a variable spectrum of clinical features, the defining features being short stature, gonadal dysgenesis and infertility.[1] Numerous autoimmune diseases in TS patients have been reported, such as juvenile rheumatoid arthritis, Addison's disease, psoriasis, celiac disease, vitiligo, alopecia areata.[2] However, SLE associated with TS has been rarely reported. Herein we present the case of a patient with TS associated with SLE.

A 24-year-old girl presented with a 10-year history of edematous erythema on the face, which was exacerbated by sunlight. She was diagnosed with TS at the age of 18 when she was referred for evaluation of short stature, amenorrhea and juvenile female external genitalia. She was 135cm in height. Butterfly facial erythema, discoid rash on the auricle, edematous/purplish erythema on the dorsum of the hands were noted (Figure 1). Secondary sex characters were lacking. Intelligence and audition were normal. No other family member had similar features. Chromosomal abnormality was confirmed by cytogenetic analysis (karyotype 45X) (Figure 2). Serum test for antinuclear antibodies (ANA) was positive at a titer of 1:160. Anti-Ro(SSA) and La(SSB) antibodies were positive. Routine reproductive hormones testing suggested estradiol<11.80pg/ml (referential range of follicular phase:19.5-144.2), follicle stimulating hormone 134.3mIU/ml (2.5-10.2), luteotropic hormone 48.7mIU/ml (1.9-125). Other laboratory tests were unremarkable. Skin biopsy was refused. The patient was diagnosed with TS with SLE, according to the revised criteria of the American Rheumatic Association (ARA) 1997 and the karyotype analysis result. She fulfilled four criteria: butterfly facial erythema, discoid rash, photosensitivity and ANA 1:160. She was treated for SLE with prednisone

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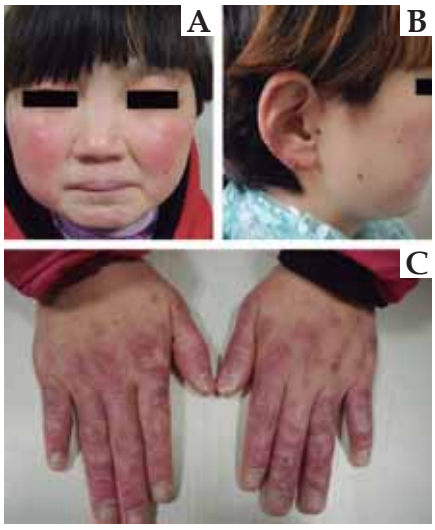

Figure 1: (a) Butterfly facial erythema, **(b)** Discoid rash on the auricle, **(c)** Edematous or purplish erythema or purplish on the dorsum of the hands

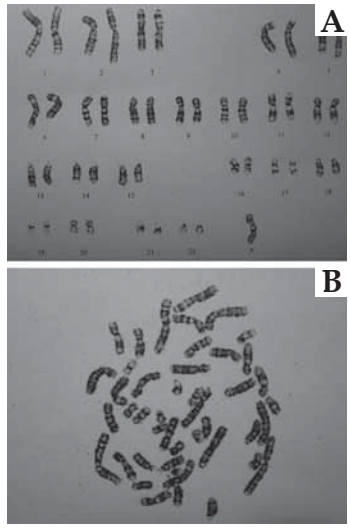

Figure 2: Photograph of the patient's chromosomal analysis of peripheral lymphocytes showing karyotype of 45X

(25mg twice daily). At the same time, she took Calcium Carbonate and Vitamin D3 Compound (calcium carbonate 1.5g vitamin D3 125IU, one tablet once daily) as a preventive measure against osteoporosis. The therapy was effective.

SLE in TS patients is extremely rare. The reported karyotypes in TS with SLE include 46,X,del(X)(q13) and 45X/46,XXq+, existing deletion of long-arm or short-arm of one X chromosome. [3,4] One such case has been reported without detailed karyotype. [5] In the current case, we reveal a novel karyotype of 45,X, completely lacking one X chromosome.

It is documented that male patients with Klinefelter's syndrome (XXY) have similar risk to develop SLE compared to females (XX). [3] This is consistent with the low prevalence of SLE in TS, characterized by an abnormal or missing X chromosome.

The pathogenetic mechanisms of SLE in TS have not been clearly elucidated. In addition to the XX chromosome, other factors may also play a significant role in the pathogenesis of SLE. Further researches are needed to shed light on the relationship between TS and SLE. ❑

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