

PHOTOCLINIC



A 13-year-old boy presented with telangiectatic erythema on his face and hemorrhagic ulcers on his lips. His medical history was significant for repeated infections, mostly of the respiratory tract.

Physical examination revealed a short stature (131 cm), low weight (20 kg), thin face, beak-like nose, telangiectatic erythema on the face, high-arched palate and hemorrhagic crusted ulcers on the lips.

A few café-au-lait spots and two hypopigmented small patches were seen on his abdomen. Histologic features of the skin biopsy specimen are shown in the Figures.

Your Diagnosis?

See next page for diagnosis

Photoclinic Diagnosis:

Bloom's Svdrome

Pathologic examination of the skin biopsy specimen showed hydropic degeneration of the basal layer of the epidermis and perivascular mononuclear infiltration in the upper dermis. Laboratory studies revealed normal results.

In 1954, Dr. David Bloom described a lupus erythematosus-like disorder of congenital telangiectatic erythema in dwarfs that subsequently became known as Bloom's syndrome. More than 150 cases have since been reported worldwide. Ashkenazi jews are among the most frequently affected population. The syndrome is inherited in an autosomal recessive manner.¹ Patients with Bloom's syndrome have a low birth weight and elongation of the head. Telangiectasis appears in light-exposed areas, particularly the cheeks, nose, malar region, and the dorsum of the hands which progress to scarring and atrophy.² Patients affected with Bloom's syndrome may have cutaneous lesions other than the telangiectasia, such as café-au-lait spots. Adjacent areas of hypopigmentation are occasionally seen.¹ These patients are particularly susceptible to infections and have low levels of immunoglobulins.²

Sexual maturation is impaired in men with Bloom's syndrome. The testes are small and most men have azoospermia. Women, in contrast, have been able to bear children. There is a slightly higher risk of mental deficiency in patients with this syndrome than in the general population but may have normal to high intellect.¹

Cultured lymphocytes and fibroblasts from patients show a high incidence of chromosomal aberrations with increased sensitivity to UV radiation. Cells with abnormally high rates of sister-chromatid exchange (SCE) are uniquely characteristic of Bloom's syndrome. The gene for Bloom's syndrome has been mapped to chromosome 15q26.1 and has been identified as a DNA helicase.³

Later in life, they are predisposed to neoplasia, particularly leukemia, lymphoma, and gastrointestinal malignancies at a higher than expected rate and at a much younger age than patients without the syndrome.^{1,2} Therefore, mortality from neoplasia during the second or third decade is significantly increased.³

References

1. Lazar AP. Answers to self-assessment examination of the American Academy of Dermatology. *J Am Acad Dermatol.* 1995; **32**: 827.
2. Arbiser JL. Genetic immunodeficiencies. Cutaneous manifestations and recent progress. *J Am Acad Dermatol.* 1995; **33**: 82-9.
3. Harper JJ. Genetics and genodermatoses. In: Rook A, Wilkinson DS, Ebling FJG, eds. *Rook/Wilkinson/Ebling Textbook of Dermatology.* Oxford, Molden: Blackwell Science; 1998: 357-436.

Reza Yaghoobi MD, Assistant Professor of Dermatology, Department of Dermatology, Ahwaz University of Medical Sciences, Ahwaz, Iran.