Case Report

A case of Behcet’s disease in a patient with albinism

Yuji NOZAKI¹, Yasuaki NAGARE², Koji KINOSHITA¹, Fumiaki URASE² and Masanori FUNAUCHI¹

¹Department of Nephrology and Rheumatology, Kinki University School of Medicine, Osaka-Sayama, Osaka, Japan,
²Department of Internal Medicine, Kinki University Sakai Hospital, Sakai, Osaka, Japan

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Summary

A 22-year-old female suffering from recurrent oral ulcers, genital ulcers, erythema nodosum, and folliculitis, was diagnosed as having Behcet’s disease (BD). She has also hypopigmentation of skin and hair, and optic changes associated with albinism including hypopigmentation of the retina, nystagmus, strabismus, and reduced visual acuity. In this report, we discuss the possibility of precipitating factor in BD that the hypersensitivity, mental stress, and drug resistance which is caused by albinism.

Key words — albino; Behcet disease

Introduction

Behcet’s disease (BD) is a multisystem disorder characterized by recurrent oral ulcers, genital ulcers, eye lesions, and skin lesions.¹

Albinism is a heterogeneous group of autosomal-recessive genetic disorders. Patients with albinism have hypopigmentation in the skin, hair and eyes, and ocular abnormalities such as reduced visual acuity and nystagmus.² This is the first report that the patients with BD and albinism.

Case report

The patient was a 22-year-old Japanese woman. At birth, she was diagnosed as having albinism based on white skin, white hair and blue eyes. She also had had optic problems associated with hypopigmentation of the retina, such as nystagmus, strabismus and reduced visual acuity, and photosensitivity dermatitis associated with hypopigmentation (Fig. 1). She had no family history of albinism.

In 2004, she noticed recurrent oral ulcers and painful erythema nodosums (EN). In December 2005, she went to another hospital because she could not control intermittent fever, polyarthritis, headache, oral ulcer, miction pain, folliculitis, and painful EN. She was senior student at the university, and she was busy at the examination and job hunting at that time. She had no history of allergic disorders and took no medicines before. Dermatologist at another hospital performed skin biopsy in her right lower thigh and the result of histopathologic findings were compatible with erythema nodosum. She was diagnosed with BD on the basis of the criteria of the International Study Group for BD ¹. Then, she had been treated with prednisolone (PSL, 15 mg/day) and colchicines, but her symptoms gradually worsened and recurrent oral ulcers, miction pain, headache, and EN became intolerable (Fig. 2). She was admitted to our hospital in January 2006. On physical examination, she had headache, oral ulcers, genital ulcers with external opening of urethra, folliculitis, EN in her right and lower thigh, and diarrhea. Laboratory tests showed an erythrocyte sedimentation rate was 48 mm/h, and white blood cell count of 8800/μl, red blood count

Fig. 1 Hypopigmentation in the skin, hair and eyes, and ocular abnormalities
cell count of $474 \times 10^4/\mu l$, platelet count of $18.2 \times 10^4 /\mu l$, and C-reactive protein level of 0.4 mg/dl. Human leukocyte antigen (HLA) B51 was positive. A slit-lamp eye examination was normal. Cerebrospinal fluid (CSF) analysis showed 4 monocytes per $\mu l$. Interleukin (IL)–6 in CSF was 0.8 pg/ml. IgG index was normal. Magnetic resonance imaging (MRI) scan of the brain was normal. There were no evidence of infections including viral and bacteria in her clinical course. These findings were seemed consistent with a diagnosis of BD (no complicated with neuro BD). However, the initial treatment PSL combined with colchicine was not sufficient to suppress her symptoms. Moreover, colchicines had to be withdrawn because of diarrhea. Therefore, 25 mg/day of PSL was further required for the improvement of BD. In a week, her symptoms gradually improved and the dose of PSL was gradually decreased. In three weeks later, she discharged our hospital and went to her university and tried to get job again. Then, her symptoms seemed better than before temporarily. However, the dosage of PSL was reduced less than 15 mg/day, her symptoms were getting worse again. Therefore we treat with the combination of PSL (15 mg) and cyclosporine (100 mg/day). In a few weeks, her symptoms were improvement gradually. However, those symptoms were not improved completely.

**Discussion**

Albinism is the term used to describe a heterogenous group of inherited disorders characterized by skin hypopigmentation and ocular abnormalities such as reduced visual acuity and nystagmus. The molecular basis of the pathogenesis in several types of albinism has been clarified since the first report in 1989 on the pathologic mutation of the tyrosinase gene.2

It may be divided broadly into two groups; oculocutaneous albinism (OCA) characterized by hypomelanosis of the hair, skin, and eyes, with autosomal recessive inheritance; and ocular albinism where skin and hair pigmentation appear to be normal.3 The molecular bases of several types of OCA have been described as follows: tyrosinase-related OCA (type I OCA), pink-eyed dilution gene-related OCA (type II OCA), tyrosinase related protein–1 OCA (type III), and unclassified.4 Type I OCA is caused by a dysfunction of the tyrosinase enzyme that catalyzes the first and second steps in the melanin synthetic pathway due to a mutation of the tyrosinase gene. Tyrosinase activity of type I OCA is completely lacking due to homogenously mutated genes of tyrosinase, and the melanin formation does not occur throughout the patient’s life. In this case, because of her family history and phenotype such as white skin and hair, blue eyes and nonpigmented
moles, we considered her albinism seems consistent with type I OCA.

The present case was regarded as BD presenting oral ulcers, genital ulcers, skin lesions and polyarthralgia. BD has higher prevalence in the countries along the ancient 'Silk Road' from Japan to the Mediterranean region.\(^5\) This patient is positive for HLA B51, which has been reported to be strongly associated with this disease in different ethnic groups,\(^6\) and its positive rate is approximately 60\%.\(^7\) There have been no reports on the patients with BD and albinism, nor evidence that suggests an association in the pathogenesis of the two diseases. Albinism may not give direct influence to the pathogenesis of BD. However, she has been exposed stressful events derived from the skin abnormality in her life and photosensitivity that are caused by albinism. In fact, at the onset of BD, she had been suffering from a depression and felt stress for her skin lesions, the examination, and job hunting. These risk factors, as well as the positivity of HLA–B51, might be trigger in the onset of BD. Once the patients with albinos would develop BD, those patients might become treatment-resistant in the view of the stress and hypopigmentation due to albinism. Further studies are needed to clarify the relationship and pathogenesis between BD and albinism.

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**References**


