

Neurofibromatosis Type 1 with Palmar Plexiform Neurofibroma and Behçet's Disease: Uncommon Association

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Abstract

Observation: We report here a rare case of Neurofibromatosis Type 1 (NF1) in a 42-year-old female who developed Behçet's disease (BD). NF1 was diagnosed twenty years ago and BD was diagnosed at 42 years of age. NF1 is an autosomal dominant disease with an incidence of 1 in 2,600 to 3,000 individuals. BD is a systemic vasculitis with obscure etiology, characterized by recurrent episodes of oral and genital ulcers, skin lesions, and ocular lesions. Although there are a few reports on the combination of Incontinentia Pigmenti and BD, this is the first reported case that concurrence of NF1 with Plexiform neurofibroma and BD.

Introduction

Behçet's disease (BD) is a systemic vasculitis with recurrence attacks. It affects many system e.g. eyes, skin, joints and also neurological involvement is possible. Rarely it accompanies to neurocutaneous syndromes. [1]. Neurofibromatosis (NF1) is an autosomal dominantly inherited neurocutaneous syndrome which prognoses with development changes on neural system, bones and skin. The most prevalent type is NF type 1 which constitutes the 85-90% of all neurofibromatosis subjects [2]. Various genetic diseases are reported to exist together with BD. These are incontinentia pigment, albinism, familial Mediterranean fever [3]. The association of NF1 and BD has not been announced yet. In this article, a case where BD accompanies NF1 and there is a giant plexiform neurofibroma in the palm, is presented.

Case Reports

A female patient aged 42, applied to our polyclinic with the complaint of ulcers in mouth and genital region. In the history of the patient, it was understood that there were maculas on her body since childhood and in the last 15 years, thickening and slouching in hand started since 15 years. Sores have been present in mouth and genital region for one month. In the dermatological examination of the patient, prevalent café-au-lait maculas, axiller freckling, giant plexiform neurofibroma in the left hand palm surface and also oral and genital ulcers were detected (Figures 1 and 2). Besides, erythematous painful nodules were present on both leg fronts. The lesions on surface of leg fronts were evaluated as erythema nodosum. Pathergy test was applied to the patient and was evaluated as negative. The laboratory values were normal. Active or old uveitis was not coincided in the medical history during eye examination. For BD, systemic colchicin 0.5 mg tb 3x1, topically chlorhexidine gluconate+benzydamine HCL gargle 3x1, diflucortolon valerate + chlorquinaldole pomade treatment



Figure 1. The genital ulcer view of the patient and prevalent café au lait maculas on the body.

was initiated for the patient. Surgical treatment was suggested for the plexiform neurofibroma but patient did not accept.

Discussion

Although etiopathogenesis of BD is not known, genetic and immune system anomalies are responsible. This disease may sometimes accompany to genodermatosis. It was also reported together with incontinentia pigment [3, 4, 5]. In another publication it was reported together with albinism [6]. Simultaneous intestinal BD was detected in maternal twins [7]. However BD and NF1 association was not reported up to now. BD presents a worldwide occurrence with varying prevalence, being endemic in the Eastern and Central Asian. A genetically determined disorder with a probable environmental triggering factor. In BD believed that an autoimmune process and a human leukocyte antigen (HLA)-B51 are the strongest associated risk factors. The etiology of the disease remains unknown, although genetic factors, infectious agents, environmental pollution, immunological mechanism have been implicated [8].

NF1 is inherited in the form of an autosomal dominant. Although a numerous of NF1 mutation have been defined, only two genotype-phenotype correlation have been noted. The gene of NF1 is located on chromosome 17q, and encodes a protein called neurofibromin. Neurofibromatosis results from a mutation of the NF1 gene [9]. Disease has same distribution between male and female, and in one half of patients there is a family history of neuro-



Figure 2. The oral aptha of the patient and plexiform neurofibroma in the left palm

fibromatosis [2]. The diagnostic features of NF1 was recognized by the National Institutes of Health in 1987. Two or more of diagnostic features is present before a diagnosis of NF1 is established [9]. Our patient had three diagnostic features, these are café au lait macules, plexiform neurofibroma and axillary freckling.

Plexiform neurofibromas (PN) usually occur in persons with NF. Cutaneous PN may occur from superficial peripheral nerves, in which condition there is no deep involvement, or can represent the superficial appendage of deeper, more massive, plexiform tumor [10].

PN involving the trunk and limbs may be related with soft tissue excessive growth, occasionally leads to serious hypertrophy. Growth of PN may arise at any time in life. Clinical experience suggests that PN tend to grow at two different periods. First is early childhood, second is during times of hormonal change, especially during puberty or, in women, during pregnancy [10, 11]. PN of our patient's was in the palm of the left hand.

BD give rise to organ damage, including the eyes, skin, joints, etc., that produces variety of clinical manifestations. The basic histopathologic characteristic is systemic vasculitis with perivascular inflammatory infiltrates. Consequently, the recent progresses in the knowledge of BD pathogenesis facilitate for innovative therapy [12].

In a study made in recent years, BD was reported to associate with familial Mediterranean fever [13]. Up to now, BD and NF1 association has not been announced and a data regarding the relation has not been repor-

ted. However as NF1 is a source of stress in the patient and stress effects immune system, it is possible that occurrence of BD may be eased. Besides genetic factors playing role in both diseases and skin and eye uptake being at the forefront, suggest that there may be an association.

Further studies are required to make clear the relationship and pathogenesis between BD and NF1.

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