GENETICS AND GENODERMATOSES

CLINICAL AND GENE MUTATION ANALYSIS OF BLAU SYNDROME

Yi Zheng [(1)] - Xinyue Zhang [(1)] - Jing Li [(1)] - Songmei Geng [(1)]

Department Of Dermatology, Northwest Hospital Affiliated To Xi'an Jiaotong University, Xi'an, China [(1)]

Introduction:Blau Syndrome(BS) is rare autoinflammatory diseases characterized by dermatitis, arthritis, uveitis and other organ systems involved with non-caseating granuloma in pathology. It has proved that mutation of NOD2/CARD15 gene lead to nuclear factor κB (NF-κB) activation contributing to BS disorders.

Objective: How to recognize the diseases in early stage and predict its prognosis is very important in clinic.

Methods: Eight cases of BS diagnosed in our hospital were analyzed on their clinical features, histopathology and gene mutation of NOD2/CARDl5.

Results: Of our 8 cases, including 4 girls and 4 boys, onset age was from 3 to 18 months. There were 3 cases had skin lesions only, 3 cases had skin lesion and arthritis, 1 case had skin lesions and uveitis, 1 case had triad symptoms of dermatitis, uveitis and hypertension, 1 case has dermatitis, uveitis and central nerve system involved; 6 cases presented generalized follicular papules and 2 cases presented erythematous plaques. There were 2 cases had intermitted fever. All skin biopsy revealed non-caseating granuloma or epithelioid granuloma in dermis. NOD2/CARD15 gene screening indicated five cases with mutation p.R334W(c.1000C>T); one case had a double mutation with a novel mutation H313R(c.938A>G) and a reported mutation R471C(c.1411C>T); one case showed negative results and one case rejected the test.

Conclusions: Early onset, multiple organs affected, and skin lesions presented with generalized follicular papules or erythematous plaques could be the signs to suspect the diagnosis of BS. Gene mutation screening of CARD15/NOD2 could help to diagnose BS and predict the prognosis.