GENETICS AND GENODERMATOSES

THE H SYNDROME: 6 NEW CASES FROM THE SOUTH OF TUNISIA

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Introduction: H syndrome is an autosomal recessive genodermatosis with multisystem involvement caused by mutations in SLC29A3.

Objectives: to assess the clinical, biological and histological features with the epidemic profile of H syndrome in south Tunisia.

Materials and Method: We conducted a retrospective descriptive clinicopathological study of 6 patients diagnosed in our dermatology department.

Results: A total of 6 patients (sex ratio: 0.5) were included in our study. The averge age was 26 years. A first-degree relative was observed in five patients. Four patients were diagnosed with congenital hearing loss. Two cases of growth restriction were noted with a dysmorphic facial features in one case. All of our patients had cutaneous hyperpigmentation, hypertrichosis and induration of the skin. The lower limbs were affected, mainly in the thighs'region in all of our 6 patients. Clinical examination revealed hepatomegaly (2cases), splenomegaly (1case), bilateral hallux valgus (5cases) and camptodactyly (5cases). Averge CRP level was 59 mg/dl and ESR was 95 mm/h. A thyroid disorder and diabetes have been detected each in 2 cases. Hyperprolactinemia was noted in 50% of cases and hypogonadism in 33 %. Only third of the cases had elevated ANA titers. Echocardiography showed cardiomegaly (1case), pericardial effusion (2cases) and ventricular wall hypertrophy (2cases). Histopathologic findings revealed epidermal basal hyperpigmentation (6cases), dermal fibrosis (6cases) and histiocytic infiltrate expressing CD 68 (2cases). The mutation of the SLC 29A3 gene in the homozygous state was isolated in 2 patients (p.R363Q and p.P324L) and is currently under study for the other patients.

Conclusion: H syndrome is a new form of systemic histiocytosis with characteristic cutaneous manifestations. The high frequency of cardiac and oesteroarticular malformations in our case series with the predominance of endocrine manifestations suggest an underlying mitochondrial disorder. The presence of an inflammatory syndrome and the positivity of AAN suggests an auto-inflammatory origin.