Clinical and Immunologic Data in Two Groups of Familial and Sporadic Patients with Common Variable Immunodeficiency

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Abstract

Background: Common variable immunodeficiency (CVID) is the most frequent symptomatic primary immunodeficiency disease and its prevalence varies significantly among different population. Minority of CVID patients present a familial aggregation suggesting a higher probability of heritable genetic defects.

Methods: A total of 235 registered CVID patients were evaluated in this cohort study. Familial and sporadic patients were stratified and demographic information, clinical records, laboratory and molecular data were compared among these two groups of patients.

Results: Multiple cases were identified in 12 families (30 patients) and sporadic presentation in 120 cases. The rate of parental consanguinity (83.3%) and clinical presentation of lymphoid malignancy (20.7%) were predominant in familial CVID patients, whereas significantly increased recurrent upper respiratory infections were recorded in sporadic patients (0.3 infections per year). Probands of familial group were presented with a higher severity score resulting in a profound mortality rate (41.7% after 30-years follow-up) comparing to the non-proband CVID patients in the same families with a lowered diagnostic delay.

Conclusion: Familial CVID patients had a specific signature in clinical presentation and immunologic profile and a high consanguinity in this group of patients suggests a Mendelian trait with an autosomal recessive inheritance pattern. Diagnosis of an index patient within a multiple case families significantly improves the diagnostic process and outcomes of the yet asymptomatic patients.

Keywords: Common variable immunodeficiency, Familial, Sporadic, Diagnosis