Clinical, Immunological, and Genetic Features of 11 Patients with Chronic Mucocutaneous Candidiasis

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Introduction:

Chronic Mucocutaneous Candidiasis (CMC) is a group of disorders that are characterized by recurrent or persistent fungal infections of the skin, mucous membranes, and nails with Candida species, often C.albicans. CMC is caused by inherited defects in the T-cell immunity specifically against Candida. Several gene mutations have been identified as causing CMC, including mutations in Signal transducer and activator of transcription 1 (STAT1), autoimmune regulator (AIRE), Caspase Recruitment Domain-containing Protein 9 (CARD9), and Interleukin-17 Receptor A/C (IL-17RA/IL-17RC) and others.

Objective:

The purpose of this study is to present the clinical manifestations and molecular findings of 11 CMC patients within 7 families who have been diagnosed with the disease.

Methods:

The study included eleven patients with suspected CMC who were referred to the Immunology, Asthma & Allergy Research Institute (IAARI) since 2008. The diagnosis was based on their clinical history, physical examination, laboratory, and immunological evaluations. To identify

genomic variants, DNA samples obtained from the whole blood of the patients were subjected to Whole exome sequencing and Sanger sequencing.

Results: Candida infections were present in all patients, and their lymphocyte transformation test (LTT) was negative. These patients did not exhibit any abnormalities in other immunological screening tests. Their genetic results revealed three novel mutations in STAT1 (c.1159 A>G), IL-17R (c.722-730 del), CARD9 (c.1032del-AGGC), as well as a previously reported 13 base pair deletion in the AIRE gene. No known variants were identified in 4 patients, and further analysis is being conducted.

Conclusion:

This study provides versatile clinical and genetic implications for CMC patients. The results of this study can contribute to the provision of genetic counseling for these families to find affected members and early diagnosis. Moreover, the development of innovative therapeutic approaches for CMC would be applicable.