SYMPOSIUM 1: Update: Primary Immune Deficiency Disorders Associated with Chronic Rhinosinusitis and Malignancy

Chronic Rhinosinusitis, Good’s Syndrome and CVID
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Primary Immune Deficiency Disorders (PIDs) Associated Malignancies and Lymphoproliferative Diseases
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Abstract

Introduction: Genetic variation, innate immune genes, and inflammatory mediators might contribute to the pathogenesis of chronic rhinosinusitis linked with immune deficiency. Good’s syndrome and common variable immune deficiency (CVID) are associated with chronic rhinosinusitis. Good’s syndrome is characterized by hypogammaglobulinemia, B-cell depletion, defects in cellular immunity and thymoma. Malignant monoclonal gammopathies and T cell tumors have been described in Good’s syndrome by several authors.

The first presenter organizer and session chairperson Marianne Frieri, M.D., Ph.D. will give a brief overview of chronic rhinosinusitis linked with immune deficiency, Good’s syndrome and CVID associated with malignancy. CVID is a heterogeneous group of disorders, which presents not only with acute and chronic infections but also with an increased incidence of lymphoma and other malignancies. The overall incidence of malignancies in CVID appears to have increased. According to several authors lymphoma was more common in females than males and of these, non-Hodgkin B-cell lymphomas were the most common, some of these being further classified into specific B-cell phenotypes, including mucosa-associated lymphoid tissue lymphoma, marginal zone lymphoma, and T cell–rich B-cell EBV-associated lymphoma. Lymphomas in CVID are usually extranodal, B cell in type, and unlike lymphomas in other congenital immune defects, are more common in subjects in the fourth to seventh decade of life and EBV negative according to several authors. In CVID patients with stage I or II disease, with non-Hodgkin lymphomas in general, the prognosis is good and several authors have stated that cytogenetic studies in lymphomas show that cytogenetic abnormalities, including chromosomal translocation, can be found in this group, but more studies will be needed to assess the frequency of these events.

The second presenter Joseph A. Bellanti, M.D will discuss the primary immune deficiency (PID) conditions. These are a group of over 200 genetic disorders that result from one or more abnormalities of 4 components of the immune system (phagocytes, complement, T lymphocytes and B lymphocytes) that manifest primarily as an increased susceptibility to infection. Because of the dysfunction of the immune system, patients with the PID disorders sometimes also present with autoimmune disease or malignancies particularly the lymphoreticular malignancies. This presentation will include a discussion of the general features of the molecular and phenotypic characteristics of the primary immunodeficiencies, the key molecular defects responsible for immunodeficiency disorders affecting various components of the innate and adaptive immune systems and a description of how the molecular defects seen in the primary immunodeficiencies contribute to the diagnosis and management of the clinical manifestations of the disorders. The presentation will also include a discussion of recently described pathogenetic mechanisms for the development of cancer and the lymphoreticular malignancies in these disorders involving a description of the key affected checkpoints which have offered unique opportunities for immunotherapeutic intervention with newly emerging biological agents.