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GILBERT OMENN AND HIS SYNDROME

GILBERT OMENN I OPISANY PRZEZ NIEGO ZESPÓŁ CHOROBOWY

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Abstract
Gilbert Omenn is a well-known American Geneticist. In the 1965, He reported a rare genetic disorder —later known as Omenn syndrome. This syndrome is a variant of severe combined immunodeficiency which is associated with hypereosinophilia. It is one of the differential diagnoses of neonatal erythroderma.
This concise report sheds light on Gilbert Omenn and the syndrome that bears his name.

Streszczenie
Gilbert Omenn jest znanym amerykańskim genetykiem. W 1965 r. opisał rzadką genetyczną chorobę, znaną później jako zespół Omenna. Ten zespół jest wariantem ciężkiego, złożonego niedoboru odporności, związanego z eozynofilią. Jest jednym z rozpoznań różnicowych noworodków erytrodermii.
Ten zwięzły raport rzuca światło na Gilberta Omenna i zespół chorobowy, który nosi jego imię

Key words: dermatology; erythroderma; Omenn syndrome
Słowa klucze: dermatologia; erytrodermia; zespół Omenna

Introduction

Gilbert Omenn (Fig. 1) is currently, a Director of Center for Computational Medicine and Bioinformatics and Professor of Internal Medicine, Human Genetics, & Public Health, at University of Michigan, Ann Arbor, MI, USA [1].
He is a world-renowned geneticist. Among his great medical contributions, he is credited for describing a reticuloendotheliosis with eosinophilia in several individuals in related sibships from an inbred American family of Irish extraction [2], later known as Omenn syndrome (OS) [3-6].

Omenn syndrome

OS (OMIM #603554) is a rare autosomal recessive genetic disorder and presents symptoms of severe combined immunodeficiency (SCID). It is characterized by erythrodermia, eosinophilia, hepatosplenomegaly, lymphadenopathy, alopecia and elevated serum IgE levels.
Similar to other patients with SCID, patients with OS present early in infancy with viral or fungal pneumonitis, chronic diarrhea, and failure to thrive. Unlike typical SCID, patients with OS have enlarged lymphoid tissue, severe erythroderma, increased IgE levels, and eosinophilia [5].

Figure 1. Gilbert Omenn

OS is also known as Reticuloendotheliosis, familial with eosinophilia, and also known as severe combined immunodeficiency with hypereosinophilia [3].
The syndrome is reported from different places in the world but large series of patients were reported from Middle East.
Some authors [6] performed literature search, for Omenn syndrome, using Medline, encompassing the period 1965-1999. They collected 68 cases and they founded that median age at onset of symptoms, in this disorder, was 4 weeks. Key symptoms were erythematous rash (98%), hepatosplenomegaly (88%), and lymphadenopathy (80%), often accompanied by recurrent infections (72%) and alopecia (57%). An elevated WBC (55%) was frequently observed, due to eosinophilia and/or lymphocytosis. B-cell counts were significantly decreased whereas T-cell counts were elevated. A high serum IgE was another frequent finding (91%). Therapeutic options, which were used in the reviewed patients, include bone marrow transplantation or cord blood stem cell transplantation; however, the mortality was 46%. The authors concluded that, Omenn syndrome is a fatal disease if untreated. They believed that, the mortality may be reduced when diagnosis is established early and treatment is initiated rapidly by using early compatible bone marrow transplantation or cord blood stem cell transplantation [6].
The syndrome can be caused by mutation in the recombination activating genes (RAG1 and RAG2) genes on chromosome 11p and the Artemis gene (DCLRE1C) on chromosome 10p [3].
In this disorder, there is a peculiar severe T-cell immune deficiency associated with autoimmunelike manifestations. Dysregulations of the central and peripheral immune tolerance, mediated by the protein autoimmune regulator (AIRE) and regulatory T cells, respectively, were proposed as possible mechanisms of this aberrant inflammatory process.
Erythroderma is one of the main features of this syndrome. The rash may be present at birth or evolve over the first few weeks of life. There is also lymphadenopathy, particularly of the axillary and inguinal nodes, as well as increased serum IgE levels with a marked eosinophilia and combined immunodeficiency [4]. Children usually present in early infancy but atypical forms may present later in the first year of life. Children usually suffer diarrhea, failure to thrive and persistent infection, as seen in other forms of SCID [4].

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The clinical picture may resemble SCID with maternofetal engraftment, when maternal T lymphocytes crossing the placenta cause a graft-versus-host disease-like reaction in an immunoincompetent patient [4].
Histology of the skin shows a dense, dermal perivascular lymphohistiocytic infiltrate, comprising activated T lymphocytes, with numerous eosinophils. S100-staining Langerhans' cells are usually absent, and there is no epidermotropism.
In the older literature this condition has been confused with Langerhans' cell histiocytosis (Letterer–Siwe disease) [4].
It is proved that both HLA-identical and haploidentical allogenic bone marrow transplant (BMT) can cure Omenn syndrome, provided that parenteral nutrition and immunosuppressive therapy are given before transplantation [3].

Gilbert Omenn

Gilbert Omenn was born 30-Aug-1941, in Chester, PA, USA. He received his education and served in several institutes in USA. He describes Omenn syndrome while he was a medical student in Harvard.
Gilbert Omenn published more than 400 papers and he authored and edited more than 15 books. He holds various scientific assignments inside and outside USA. He is active in many cultural and educational organizations. He has won several honors and awards.
This short report can not encompass his impressive career which can be read online in many internet websites [1]. Music and Tennis is among Dr Omenn extra scientific interests.
His research interests include clinical informatics, databases and computing, medical and translational research, and proteomics [1].
Dr. Omenn's research focuses on cancer proteomics and informatics. He leads the Proteomics Alliance for Cancer Research, the HUPO Plasma Proteome Project, the Driving Biological Problems Core of the National Center for Integrative Biomedical Informatics, and the Center for Computational Medicine and Bioinformatics [1].
He has long-standing interests in mechanisms of genetic predispositions to risks from environmental and occupational exposures, pharmacogenetics and pharmacogenomics, and science-based risk analyses [1]. He was, also, the principal investigator for many years of the Beta-Carotene and Retinol Efficacy Trial (CARET) for chemoprevention of lung cancers and heart disease.

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