Allergol Immunopathol (Madr). 2016;44(4):322–330

Allergologia et immunopathologia

Sociedad Española de Inmunología Clínica, Alergología y Asma Pediátrica

www.elsevier.es/ai

ORIGINAL ARTICLE

Long-term follow-up of ninety eight Iranian patients with primary immune deficiency in a single tertiary centre

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Received 4 May 2015; accepted 30 September 2015
Available online 20 January 2016

KEYWORDS
Consanguinity;
Complications;
Follow-up;

Abstract
Purpose: The aim was to describe the clinical manifestations, complications and long-term outcome of a cohort of Iranian patients with primary immune deficiency (PID).

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http://dx.doi.org/10.1016/j.aller.2015.09.006

Iran;
Manifestations;
Mortality;
Primary immune deficiency

Method We retrospectively studied the demographic, clinical and immunological characteristics of the PID patients in a single tertiary centre, from January 1989 to July 2014. The patients were classified according to the International Union of Immunological Societies Expert Committee on PID.

Results: 98 patients were diagnosed with and followed-up for 15 disorders. The mean age at onset and diagnosis and the diagnostic delay were 8 ± 10, 14.2 ± 13.1 and 6.1 ± 7 years, respectively. Parental consanguinity rate was 57%. Predominantly Antibody Deficiency was the most common diagnosis ($n = 63$), followed by congenital defects of phagocytes ($n = 16$), combined immunodeficiencies ($n = 12$), well defined syndromes ($n = 4$) and defects in innate immunity ($n = 3$). Recurrent sinopulmonary infection was the most common presentation. Active infections were treated appropriately, in addition to prophylactic therapy with IVIG and antimicrobials. Not all the patients were compliant with prophylactic regimens due to cost and unavailability. One SCID patient underwent successful bone marrow transplantation. The total mortality rate was 19% during the follow-up period (7.8 ± 7.6 years). The mean age of living patients at the time of study was 23 ± 11.7 years.

Conclusions: Physicians awareness of PID has been rising dramatically in Iran, ensuring an increasing number of patients being diagnosed and treated. More effective treatment services, including health insurance coverage and drug availability are needed to improve the outcome of PID patients.

Introduction

Primary immunodeficiency disorders (PID) are rare inherited diseases of the immune system that present with heterogeneous infectious and autoimmune manifestations and higher incidence of malignancies.[1–6] The International Union of Immunological Societies (IUIS) Expert Committee classified PID in nine main groups.[7] Overall, the prevalence of PID is estimated to be one in 1200 American people and appears to be underdiagnosed.[2] The aim of this study was to summarise the clinical manifestations and long-term outcome of 98 Iranian PID patients in a single tertiary centre.

Method

This was a single-centre retrospective study carried out at Allergy and Clinical Immunology Department of Rasool-E-Akram, an 830-bed tertiary referral hospital, in Tehran, the capital city of Iran, from January 1989 to July 2014 (Fig. 1).

The diagnosis of PID was made after secondary forms of immunodeficiency were ruled out and according to clinical and laboratory findings and diagnostic criteria of IUIS, PAGID (Pan-American Group for Immunodeficiency) and ESID (European Society for Immunodeficiencies).[1,7,8] Genetic testing (to confirm the diagnosis of X linked agammaglobulinaemia, X linked hyper IgM, LRBA deficiency, DOCK8 deficiency, STAT1 and HAX1) was done as part of genetic consultation with Research Center for Immunodeficiency in Children's Medical Center in Tehran University of Medical Sciences. Only patients with a definite diagnosis of PID were enrolled for the purpose of the study. Demographic, clinical and immunological data were collected using a standardised questionnaire.

Serological and immunological laboratory testing included quantitative serum immunoglobulin levels, IgG subclass levels, pre- and post-immunisation antibody titres to Pneumococcus, Diphtheria and Tetanus, isohaemagglutinin titres, flow cytometric assessment of lymphocyte subsets, lymphocyte transformation test, nitroblue tetrazolium test, complement component (C3, C4, CH50) tests and delayed type hypersensitivity skin testing to Candida antigen.

The majority of the patients were diagnosed at Rasool-E-Akram Hospital, whereas some of the older patients

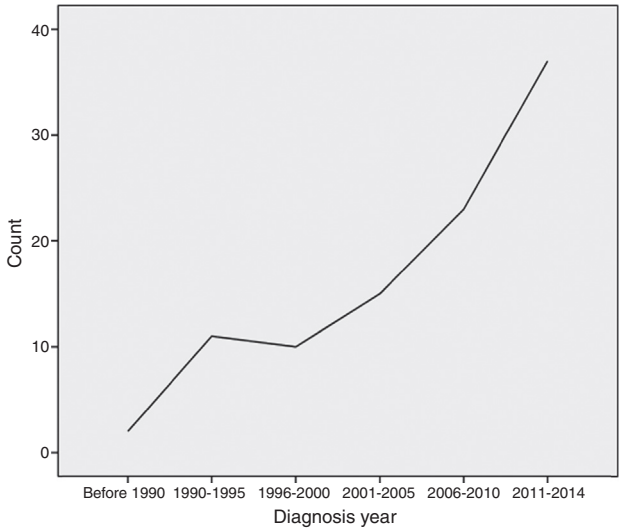

Figure 1 Number of newly diagnosed patients with primary immune deficiency at Rasool-E-Akram Hospital, Iran University of Medical Sciences (IUMS), Tehran, Iran between 1990 and 2014.