



ORIGINAL ARTICLE

Doctors' awareness concerning primary immunodeficiencies in Brazil



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Abstract

Background: PIDs are a heterogeneous group of genetic illnesses, and delay in their diagnosis is thought to be caused by a lack of awareness among physicians concerning PIDs. The latter is what we aimed to evaluate in Brazil.

Methods: Physicians working at general hospitals all over the country were asked to complete a 14-item questionnaire. One of the questions described 25 clinical situations that could be associated with PIDs and a score was created based on percentages of appropriate answers.

Results: A total of 4026 physicians participated in the study: 1628 paediatricians (40.4%), 1436 clinicians (35.7%), and 962 surgeons (23.9%). About 67% of the physicians had learned about PIDs in medical school or residency training, 84.6% evaluated patients who frequently took antibiotics, but only 40.3% of them participated in the immunological evaluation of these patients. Seventy-seven percent of the participating physicians were not familiar with the warning signs for PIDs. The mean score of correct answers for the 25 clinical situations was 48.08% (± 16.06). Only 18.3% of the paediatricians, 7.4% of the clinicians, and 5.8% of the surgeons answered at least 2/3 of these situations appropriately.

Conclusions: There is a lack of medical awareness concerning PIDs, even among paediatricians, who have been targeted with PID educational programmes in recent years in Brazil. An increase in awareness with regard to these disorders within the medical community is an important step towards improving recognition and treatment of PIDs.

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Introduction

PIDs encompass more than 200 genetic illnesses, predispose patients to infections, allergy, and autoimmune and malignant manifestations and are caused by immune system defects.¹ Individual PIDs are considered rare diseases, while as a group, the estimated frequency ranges from 1:2000 to 1:10,000.^{2,3} However, many of these diseases have not yet been completely described.⁴

Over the last few decades, advances in immune system studies have allowed for a greater understanding of PID pathophysiology, which has enabled greater diagnostic precision as well as recommendations of new therapeutic strategies.⁵ On the other hand, the great diversity of clinical and genetic defects makes recognition and diagnosis of patients with PID a challenge.⁶

Severe defects are easily recognisable, but there are cases in which PID is diagnosed only when the patient has been hospitalised one or more times and may already present permanent damage.^{7,8}

Lack of awareness among physicians concerning PIDs is reported in many countries to be the probable cause of delay in diagnosis and undiagnosed PIDs, which are generally not included in the differential diagnoses of potentially affected patients.^{9–11}

There are limited published data about physicians' awareness concerning PIDs and most of them involved only paediatricians.^{12–15}

The aim of this study is to evaluate physicians' awareness concerning PIDs in Brazil.

Methods

The study was approved by the Clinical Research and Ethics Committee of the Federal University of São Paulo – UNIFESP.

The study was cross-sectional and was conducted in general hospitals (public and private) in 27 cities throughout the country between July 2008 and January 2010. Hospitals were chosen taking into consideration the availability of medical,

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