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SERIES: PRIMARY IMMUNODEFICIENCY DISEASES IN LATIN AMERICA: PROCEEDINGS OF THE LATIN AMERICAN SOCIETY FOR IMMUNODEFICIENCIES (LASID) ADVISORY BOARD

Critical issues and needs in management of primary immunodeficiency diseases in Latin America

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Abstract Experts from six Latin American countries met to discuss critical issues and needs in the diagnosis and management of primary immunodeficiency diseases (PIDD). The diagnosis of PIDD is generally made following referral to an immunology centre located in a major city, but many paediatricians and general practitioners are not sufficiently trained to suspect PIDD in the first place. Access to laboratory testing is generally limited, and only some screening tests are typically covered by government health programmes. Specialised diagnostic tests are generally not reimbursed. Access to treatment varies by country reflecting differences in healthcare systems and reimbursement policies. An online PIDD Registry Programme for Latin America has been available since 2009, which will provide information about PIDD epidemiology in the region. Additional collaboration across countries appears feasible in at least two areas: a laboratory network to facilitate the diagnosis of PIDD, and educational programmes to improve PIDD awareness. In total, these collaborations should make it possible to advance the diagnosis and management of PIDD in Latin America.

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Introduction

Primary immunodeficiency diseases (PIDD) include almost 200 types of inherited disorders with impaired host defenses and other immune dysfunctions.[1,2] PIDD predispose affected individuals to recurrent infections, but, depending on the immune deficit, may also lead to the development of autoimmune diseases, inflammation, and malignancies.[3,4] The incidence of these diseases varies considerably from approximately 1 in 250 for the more common forms to 1 in 1,000,000 for rare types. The actual frequency of PIDD is unknown, but some estimates range from 1:10,000[3] to as many as 1:1200.[5] The most common forms of PIDD involve antibody deficiencies, whereas most remaining cases are mainly caused by cellular, combined, or phagocytic defects.[3,6] In 2007, the second report of the Latin American Group for Immunodeficiencies (LAGID) described the characteristics of 3321 PIDD patients from 12 Latin American countries with the most common category of PIDD (predominantly antibody deficiencies), accounting for 53.2% of the cases.[7] This category was followed by other well-defined immunodeficiency syndromes, such as DNA repair defects, hyper-IgE syndrome, and thymic defects (22.6%), combined T- and B-cell immunodeficiencies (9.5%), phagocytic disorders (8.6%), immune dysregulation (3.3%), and complement deficiencies (2.8%). An earlier report from the same group also concluded that antibody deficiencies account for at least half of the PIDD cases in Latin America.[8] An online PIDD Registry Programme for Latin America (the Latin American Society of Immunodeficiencies [LASID] registry) — available at http://deficiencia.unicamp.br:8080/ — was initiated in 2009, which will provide further information about PIDD epidemiology in the region.

Significant advances have been made over the last 15 years in the diagnosis and management of PIDD, resulting in reduced rates of morbidity and mortality.[9,10] North American and European guidelines have been published providing recommendations on how patients with suspected PIDD should be evaluated, which tests should be conducted to reach a specific diagnosis, and how patients should be treated depending on the type and severity of PIDD.[3,11] Nevertheless, awareness about PIDD and access to appropriate treatment may be restricted in some regions of the world. On October 14, 2009, a group of PIDD experts from six Latin American countries (Argentina, Brazil, Chile, Colombia, Honduras, and México) and representatives from LASID met in Cartagena de Indias, Colombia to discuss critical issues and needs related to each country separately and to the regions as a whole. This report provides a summary of these discussions.

Current status and issues in PIDD diagnosis

The diagnosis of PIDD is generally made after patients are referred to an immunology centre with specific expertise in these disorders. Most centres are located in major cities of each country (Table 1). Unfortunately, many paediatricians and general practitioners, as well as specialists in areas related to these disorders, are not sufficiently trained to suspect PIDD in the first place. This represents a critical issue in each country, because it delays the diagnosis and leaves many patients without appropriate treatment, both of which contribute to PIDD-associated morbidity and mortality. The Jeffrey Modell Foundation has recently shown that physician education and public awareness might significantly impact the early diagnosis of PIDD;[12] therefore, physicians educated to recognise at least the warning signs for PIDD can start to suspect PIDD and refer patients to immunologists for appropriate follow-up. The role of the immunologist is to confirm the diagnosis and indicate the most appropriate treatment and follow up. Therefore, referral to an immunology centre represents a critical step in the diagnostic process, and highlights the need to increase awareness about PIDD among Latin American physicians.

Table 1 Location of immunology centres for supporting a PIDD diagnosis.

Country	Locations
Argentina	Buenos Aires,[a,b] La Plata, Rosario, Cordoba, Mendoza
Brazil	São Paulo,[a] Ribeirão Preto,[a] Campinas,[a] Belo Horizonte, Curitiba, Porto Alegre,
Chile	Santiago, Temuco, Valparaíso, Concepción
Colombia	Medellin,[a] Bogotá, Cali
Honduras	Tegucigalpa, San Pedro Sula
México	México City,[a] Monterrey, Guadalajara

[a] Sites supporting molecular diagnosis.
[b] Of the five centres in Buenos Aires, only one is for adult patients.

Several immunology centres in Argentina can support a diagnosis of PIDD, most of which are located in Buenos Aires. Two of the five centres in Buenos Aires — Hospital de Pediatría ''Prof Dr Juan Garrahan'' and Hospital de Niños Ricardo Gutiérrez — have comprehensive immunology programmes with state-of-the-art facilities for immunological and molecular diagnosis. Other hospitals located in La Plata (Sor Ludovica), Rosario, Cordoba, and Mendoza can also support a PIDD diagnosis, but none have well-equipped facilities for molecular diagnosis. Moreover, only one of them, Hospital Durand in Buenos Aires, treats adult patients. Patients who are referred to an immunology centre in Argentina will be seen by trained immunologists and receive treatment, if necessary. In general, these centres are accessible and without cost to patients providing they are referred by a physician. Private hospitals provide only a partial solution, because they lack immunology laboratories and trained immunologists to support a PIDD diagnosis.

In Brazil, the diagnosis of PIDD is supported by immunology centres in a few large cities, including São Paulo, Ribeirão Preto, Campinas, Belo Horizonte, Curitiba, Porto Alegre, Salvador, and Rio de Janeiro. Of these, São Paulo, Ribeirão Preto, and Campinas have university centres qualified for comprehensive laboratory testing, including molecular diagnosis. Approximately 10 other centres, also located in large cities, are able to recognise patients with PIDD but only have the facilities to do basic lab exams and thus need support from São Paulo. The government sponsors a programme for patients who do not have specialists in their hometown that pays for travel to another city for diagnosis. However, patients living in some regions of the