Abstract Code: ASID021-019

First report on the implementation of primary immunodeficiencies (PID) biological diagnosis in Senegal: diagnostic challenges

MS. Guèye[1], I. Dème/Ly[2], A. Kane[2], A. Nakoulima[3], MA. Diouf[4], M. Ndour[1], AA. Diallo[1], R. Seck/Diao[5], M. Gadji[5], B. Mbengue[1], TN. Dièye[5]
sokho1209nana@gmail.com
Senegal

[1] *Laboratoire d'Immunologie Hôpital Aristide le Dantec*
[2] *Centre Hospitalier National d'Enfants Albert Royer (CHNEAR) Dakar Sénégal*
[3] *Service de Pédiatrie Hôpital Principal de Dakar Sénégal*
[4] *Centre hospitalier de Pikine*
[5] *Centre National de transfusion sanguine.*

Keywords: primary immunodeficiency, cytometry, electrophoresis.

Abstract

Introduction: The primary immunodeficiencies (PID) are heterogeneous groups of more than 330 genetic diseases. They are a large group of diseases affecting the immune system, predisposing the patients to a large spectrum of infections.

Aim: Conduct a biological exploration of PID with the available technical means to reinforce an established clinical diagnosis.

Methods: From August 2014 to December 2020, we have retained the suspected cases of PID from several hospital-university structures in Dakar. The included patients were referred to the laboratory of the National Blood Transfusion Centre for blood count and electrophoresis of proteins; and the Immunology service of Dantec hospital for immunophenotyping.

Results: We registered 33 patients, with a sex ratio (M/F = 0.73) and an average age of 6.10 years (extremes: 0-42 years). All the patients have a CBC: neutropenia was found in 8 patients, lymphopenia in 6 and 8 thrombocytopenia. For immunotyping: a decrease in CD4+ cells was observed in 8 patients, 4 patients have a decrease of lymphocytes CD8+ and 8 for B cells. Protein electrophoresis showed inflammatory profile in 81.25% of cases. The weighting of immunoglobulin, complement examination and the only one genetic confirmation were done in specialized laboratory. Among other, we obtained 6 cases of syndromic combined immune deficiencies, 4 SCID patients, 4 had humoral deficiency and 2 cases of verruciform epidermodysplasia.

Conclusion: PIDs are rare diseases but not exceptional. Diagnosis is a strong clinical suspicion, with biological orientation, but confirmation is genetic in several cases.