First experience in Paraguay on genotypic characterization of pediatric patients with inborn errors of immunity

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ABSTRACT

Inborn errors of immunity, also called primary immunodeficiencies (PID), are pathologies with low suspicion and underdiagnosed, mainly in countries with limited access to their diagnostic tests. The aim of this study was to genotypically characterize twelve pediatric patients who had a previous phenotypic (clinical-immunological) diagnosis of PID. Between March and May 2022, buccal swab samples were taken at the Instituto de Investigaciones en Ciencias de la Salud-Universidad Nacional de Asunción-Paraguay, and sent to Invitae Laboratory-California USA, under the sponsorship of Jeffrey Modell Insights Program. Next-generation sequencing for a panel of 429 PID genes was performed. Nine of the genotypic detections agreed with the previous phenotypic diagnoses: 2 cases of BTK mutation (Exons 8-10 deletions and c.1793A>G) of the X-linked agammaglobulinemia, 2 cases of CYBB mutation (c.469C>T and c.866G>A) of X-linked chronic granulomatous disease (CGD), one autosomal recessive CGD (NCF2 c.338G>A, a novel mutation not published yet), 2 hyper IgM syndromes (both brothers with the AICDA mutation c.93C>A), 2 Wiskott-Aldrich syndromes (WAS mutation c.264C>A and c.361-1G>C splice acceptor). However, in three patients, the genotypic detection did not coincide with the phenotype: Di-George syndrome (TBX1 gene deletion-entire coding sequence), C2 deficiency (c.841_849+19del splice site), X-linked lymphoproliferative syndrome (XIAP mutation c.1141C>T), being that these patients had a previous phenotypic diagnosis of antibody deficiencies, thus they were reevaluated by a specialized medical team to make the most appropriate diagnostic and therapeutic decisions. In most cases of PID the phenotypic diagnosis is sufficient for the patient's appropriate treatment, which is very important in countries like Paraguay or others of our region where the access to diagnostic tests is limited. However, there are cases where the genotypic diagnosis is necessary to specify the treatment, in which case the sponsorship by international agents is crucial mainly due to high costs

BIOGRAPHY

Diana Sanabria completed her studies in Biochemistry at the Universidad Nacional de Asunción-Paraguay, also finished a Master's degree in Biomedical Sciences, and she is specialized in Research Methodology and University Higher Education. She works as a researcher at the Universidad Nacional de Asunción in Clinical Immunology area, specifically on primary immunodeficiencies/inborn errors of immunity in pediatric patients. The research group that she leads is the only one in Paraguay that focused in these pathologies and publishes articles at the local level.

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